Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MENDELIAN INHERITED DISORDERS PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.1.0 (5566 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
A2M	96.1%	96%	100%	100%	99.8%	
A4GALT	100%	100%	100%	100%	98.8%	[Blood group, P1Pk system, P(2) phenotype], 111400;NOR polyagglutination syndrome, 111400;[Blood group, P1Pk system, p phenotype], 111400
AAAS	100%	100%	100%	100%	99.7%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	90.7%	90.7%	100%	100%	99.8%	Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AASS	100%	100%	100%	100%	99.8%	Hyperlysinemia, 238700
ABAT	100%	100%	100%	100%	99.4%	GABA-transaminase deficiency, 613163
ABCA1	100%	100%	100%	100%	99.8%	Tangier disease, 205400;HDL deficiency, familial, 1, 604091
ABCA12	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCA2	100%	100%	100%	99.9%	98.7%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCA3	97.2%	97.2%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 3, 610921
ABCA4	100%	100%	100%	100%	99.7%	Retinal dystrophy, early-onset severe, 248200;Retinitis pigmentosa 19, 601718;{Macular degeneration, age-related, 2}, 153800;Cone-rod dystrophy 3, 604116;Fundus flavimaculatus, 248200;Stargardt disease 1, 248200
ABCA5	100%	100%	100%	100%	99.8%	?Hypertrichosis, congenital generalized, with gingival hyperplasia, 135400
ABCB1	100%	100%	100%	100%	99.7%	Encephalopathy, acute transient, 620950;{Inflammatory bowel disease 13}, 612244;{Colchicine resistance}, 120080

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ABCB11	100%	100%	100%	100%	99.7%	Cholestasis, benign recurrent intrahepatic, 2, 605479;Cholestasis, progressive familial intrahepatic 2, 601847
ABCB4	100%	100%	100%	100%	99.8%	Gallbladder disease 1, 600803;Cholestasis, intrahepatic, of pregnancy, 3, 614972;Cholestasis, progressive familial intrahepatic 3, 602347
ABCB6	100%	100%	100%	100%	99.5%	Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153;Microphthalmia/coloboma 7, 614497
ABCB7	100%	100%	99.6%	92.7%	74.7%	Anemia, sideroblastic, with ataxia, 301310
ABCC1	96.8%	96.8%	100%	100%	99.4%	?Deafness, autosomal dominant 77, 618915
ABCC2	100%	100%	100%	100%	99.8%	Dubin-Johnson syndrome, 237500
ABCC6	94.4%	94.4%	100%	100%	99.5%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ABCC8	100%	100%	100%	100%	99.5%	Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	96%	96%	100%	100%	99.8%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABCD2	100%	100%	100%	100%	99.7%	
ABCD3	100%	100%	100%	100%	99.5%	?Bile acid synthesis defect, congenital, 5, 616278;Oculopharyngodistal myopathy 5, 621446
ABCD4	100%	99.6%	100%	99.9%	99.3%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABCG5	100%	100%	100%	100%	99.5%	Sitosterolemia 2, 618666
ABCG8	100%	100%	100%	100%	99.7%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD16A	97.5%	97.5%	100%	100%	99.4%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100%	100%	100%	100%	99.9%	Chanarin-Dorfman syndrome, 275630
ABL1	100%	100%	100%	100%	99.4%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACACA	100%	100%	100%	100%	99.7%	Acetyl-CoA carboxylase deficiency, 613933
ACAD8	100%	99.9%	100%	99.9%	99.5%	Isobutyryl-CoA dehydrogenase deficiency, 611283
ACAD9	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADM	93.1%	91.3%	100%	100%	99.7%	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450
ACADS	100%	100%	100%	100%	99.5%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100%	100%	100%	99.9%	99.7%	2-methylbutyrylglycinuria, 610006
ACADVL	100%	100%	100%	100%	99.3%	VLCAD deficiency, 201475

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ACAN	99.1%	98.9%	98.5%	97.5%	96.1%	?Spondyloepiphyseal dysplasia, Kimberley type, 608361;Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans, 165800;Spondyloepimetaphyseal dysplasia, aggrecan type, 612813
ACAT1	100%	100%	100%	100%	99.6%	Alpha-methylacetoacetic aciduria, 203750
ACAT2	100%	100%	100%	100%	99.9%	?ACAT2 deficiency, 614055
ACBD5	85.6%	85.6%	100%	100%	99.5%	Retinal dystrophy with leukodystrophy, 618863
ACBD6	79.5%	79.5%	100%	99.9%	99.6%	Neurodevelopmental disorder with progressive movement abnormalities, 620785
ACD	100%	100%	100%	99.9%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ACE	100%	100%	100%	99.8%	98.5%	Renal tubular dysgenesis, 267430;{Microvascular complications of diabetes 3}, 612624;[Angiotensin I-converting enzyme, benign serum increase], 621420
ACER3	96.3%	96.3%	100%	100%	99.9%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACKR3	100%	100%	100%	99.9%	98.9%	?Oculomotor-abducens synkinesis, 619215

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	97.9%	97.9%	100%	100%	99.7%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACOX2	100%	100%	100%	100%	99.7%	Bile acid synthesis defect, congenital, 6, 617308
ACP4	100%	100%	100%	99.9%	99.4%	Amelogenesis imperfecta, type IJ, 617297
ACP5	100%	100%	100%	100%	99.6%	Spondyloenchondrodysplasia with immune dysregulation, 607944
ACR	100%	100%	99.5%	96.3%	90.5%	?Spermatogenic failure 87, 620500
ACSF3	94.7%	94.7%	100%	100%	99.3%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100%	100%	99.1%	91.5%	73.4%	Intellectual developmental disorder, X-linked 63, 300387
ACSL5	100%	100%	100%	100%	99.6%	?Diarrhea 13, 620357
ACSL6	100%	100%	100%	100%	99.6%	
ACTA1	100%	100%	100%	99.8%	97.9%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ACTA2	100%	100%	100%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ACTB	100%	100%	100%	99.9%	99.4%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTC1	90.7%	90.6%	100%	100%	99.6%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTG1	100%	100%	100%	99.9%	98.9%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACTG2	93%	93%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5, 619431;Visceral myopathy 1, 155310
ACTL6A	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ACTL6B	95.9%	94.6%	100%	100%	99.4%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACTL7A	100%	100%	100%	100%	99.9%	Spermatogenic failure 86, 620499
ACTL9	100%	100%	100%	100%	99.4%	Spermatogenic failure 53, 619258
ACTN1	95.9%	95.9%	100%	100%	99.4%	Bleeding disorder, platelet-type, 15, 615193
ACTN2	99.8%	98.4%	100%	99.8%	98.6%	Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158
ACTN4	93%	92.9%	100%	99.9%	99.1%	Glomerulosclerosis, focal segmental, 1, 603278
ACTRT1	100%	100%	99.3%	93.1%	73.3%	
ACTRT3	100%	100%	99.8%	98.1%	94%	
ACVR1	100%	100%	100%	100%	99.7%	Fibrodysplasia ossificans progressiva, 135100
ACVR1B	100%	100%	100%	99.9%	99.3%	Pancreatic cancer, somatic, 260350
ACVR2B	100%	100%	100%	100%	99.4%	Heterotaxy, visceral, 4, autosomal, 613751
ACVRL1	91.2%	91.2%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ACY1	100%	100%	100%	99.9%	99.4%	Aminoacylase 1 deficiency, 609924

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ADA	85.1%	84.4%	100%	99.9%	99.5%	Adenosine deaminase deficiency, partial, 102700;Severe combined immunodeficiency due to ADA deficiency, 102700
ADA2	95.1%	93.2%	100%	100%	99.8%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAD2	100%	100%	100%	100%	99.2%	
ADAM10	89.8%	89.8%	100%	100%	99.6%	{Alzheimer disease 18, susceptibility to}, 615590;Reticulate acropigmentation of Kitamura, 615537
ADAM17	96.1%	96.1%	100%	100%	99.8%	?Inflammatory skin and bowel disease, neonatal, 1, 614328;?Hypotichosis 16, 621490
ADAM22	98.7%	98.7%	100%	100%	99.7%	Developmental and epileptic encephalopathy 61, 617933
ADAM9	95%	95%	100%	100%	99.6%	Cone-rod dystrophy 9, 612775
ADAMTS1	100%	100%	100%	100%	99.7%	
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ADAMTS15	100%	100%	100%	99.9%	99%	Arthrogryposis, distal, type 12, 620545
ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ADAMTS18	100%	100%	100%	100%	99.7%	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458
ADAMTS19	94%	94%	100%	99.9%	99.3%	Cardiac valvular dysplasia 2, 620067
ADAMTS2	97.9%	97.9%	100%	99.9%	99.5%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	100%	100%	99.8%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTS9	100%	100%	100%	99.8%	98.8%	
ADAMTSL2	100%	100%	100%	99.9%	99.4%	Geleophysic dysplasia 1, 231050
ADAMTSL4	100%	100%	100%	100%	99.4%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100
ADAR	100%	100%	100%	100%	99.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADARB1	94.8%	94.7%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286
ADCK2	86.1%	86.1%	100%	100%	98.9%	
ADCK5	93.5%	93.5%	100%	100%	99.5%	
ADCY1	99.2%	98.6%	100%	99.9%	98.8%	?Deafness, autosomal recessive 44, 610154

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ADCY10	100%	100%	100%	100%	99.9%	{Hypercalciuria, absorptive, susceptibility to}, 143870
ADCY3	100%	100%	100%	100%	99.5%	{Obesity, susceptibility to, BMIQ19}, 617885
ADCY5	97.5%	96.9%	100%	99.9%	99.1%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADCY6	100%	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 8, 616287
ADD1	100%	100%	100%	100%	99.6%	{Hypertension, essential, salt-sensitive}, 145500
ADD3	97.6%	97.4%	100%	100%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGB	100%	100%	100%	99.9%	99.5%	
ADGRE2	99.9%	99.4%	99.6%	98.9%	97.8%	Vibratory urticaria, 125630
ADGRG1	100%	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854
ADGRG2	100%	100%	99.3%	91.3%	72.5%	Congenital bilateral absence of vas deferens, X-linked, 300985

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ADGRG6	100%	100%	100%	100%	99.7%	Lethal congenital contracture syndrome 9, 616503
ADGRL1	100%	100%	100%	100%	99.2%	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065
ADGRV1	100%	100%	100%	100%	99.8%	Usher syndrome, type 2C, 605472;Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472;?Febrile seizures, familial, 4, 604352
ADH5	100%	100%	100%	100%	99.6%	AMED syndrome, digenic, 619151
ADIPOQ	100%	100%	100%	100%	99.6%	Adiponectin deficiency, 612556
ADIPOR1	100%	100%	100%	100%	99.8%	
ADK	88%	88%	100%	100%	99.8%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	100%	100%	100%	100%	99.7%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADRA2A	100%	100%	100%	99.6%	96.5%	?Lipodystrophy, familial partial, type 8, 620679
ADRB1	100%	100%	100%	100%	98.9%	?[Short sleep, familial natural, 2], 618591;[Resting heart rate], 607276
ADRB2	100%	100%	100%	100%	99.7%	
ADSL	99.5%	97.5%	100%	100%	99.7%	Adenylosuccinase deficiency, 103050
ADSS1	100%	100%	100%	99.9%	99%	Myopathy, distal, 5, 617030

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AEBP1	100%	100%	100%	99.9%	99.3%	Ehlers-Danlos syndrome, classic-like, 2, 618000
AFF2	100%	100%	99.4%	90.5%	71.8%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100%	100%	100%	100%	99.4%	KINSSHIP syndrome, 619297
AFF4	100%	100%	100%	100%	99.9%	CHOPS syndrome, 616368
AFG2A	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AFP	100%	100%	100%	100%	99.8%	[Hereditary persistence of alpha-fetoprotein], 615970;Alpha-fetoprotein deficiency, 615969
AGA	91.6%	91.6%	100%	100%	99.5%	Aspartylglucosaminuria, 208400
AGAP1	100%	100%	100%	99.8%	98.5%	
AGBL1	100%	100%	100%	100%	99.8%	Corneal dystrophy, Fuchs endothelial, 8, 615523
AGBL5	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 75, 617023

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AGK	91.6%	88.9%	100%	100%	100%	Cataract 38, autosomal recessive, 614691;Sengers syndrome, 212350
AGL	100%	100%	100%	100%	99.7%	Glycogen storage disease IIIa, 232400;Glycogen storage disease IIIb, 232400
AGMO	100%	100%	100%	100%	99.7%	
AGO1	99.4%	98.4%	100%	100%	99.7%	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292
AGO2	100%	100%	100%	100%	99.6%	Lessel-Kreienkamp syndrome, 619149
AGPAT2	100%	99.9%	100%	100%	98.9%	Lipodystrophy, congenital generalized, type 1, 608594
AGPS	97.3%	97.3%	100%	100%	99.8%	Rhizomelic chondrodysplasia punctata, type 3, 600121
AGR2	100%	100%	100%	100%	99%	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233
AGRN	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120
AGT	95.7%	95.3%	100%	100%	99.5%	Renal tubular dysgenesis, 267430
AGTPBP1	96.5%	96.5%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AGTR1	100%	100%	100%	100%	99.9%	{Hypertension, essential}, 145500;Renal tubular dysgenesis, 267430

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AGXT	97.4%	96.4%	100%	100%	99.5%	Hyperoxaluria, primary, type 1, 259900
AHCY	100%	100%	100%	99.9%	99.1%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100%	100%	100%	100%	98.9%	Xia-Gibbs syndrome, 615829
AHI1	98.7%	98.7%	100%	99.9%	99.6%	Joubert syndrome 3, 608629
AHNAK2	97.8%	97.7%	95.6%	94.1%	91.1%	
AHR	100%	100%	100%	100%	99.8%	Foveal hypoplasia 3, 620958;?Retinitis pigmentosa 85, 618345
AHSG	100%	100%	100%	100%	99.8%	?Alopecia-intellectual disability syndrome 1, 203650
AICDA	92.1%	92.1%	100%	100%	99.7%	Immunodeficiency with hyper-IgM, type 2, 605258
AIFM1	100%	100%	99.3%	90.3%	72.6%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AIMP1	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100%	100%	100%	99.8%	99.1%	Leukodystrophy, hypomyelinating, 17, 618006
AIP	100%	99.5%	100%	100%	99.8%	Pituitary adenoma 1, multiple types, 102200;Pituitary adenoma predisposition, 102200

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AIPL1	100%	100%	100%	99.9%	98.9%	Leber congenital amaurosis 4, 604393;Retinitis pigmentosa, juvenile, 604393;Cone-rod dystrophy, 604393
AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AJAP1	100%	100%	100%	100%	99.1%	
AK1	100%	100%	100%	100%	99.8%	Anemia, congenital, nonspherocytic hemolytic, 3, adenylate kinase deficient, 612631
AK2	98.1%	93.2%	100%	100%	99.1%	Reticular dysgenesis, 267500
AK3	100%	100%	100%	100%	99.9%	
AK7	97%	96.1%	100%	100%	99.7%	?Spermatogenic failure 27, 617965
AK9	100%	100%	100%	100%	99.9%	Spermatogenic failure 89, 620705
AKAP3	100%	100%	100%	100%	99.9%	Spermatogenic failure 82, 620353
AKAP4	100%	100%	98.9%	91.3%	73.8%	
AKAP9	99.6%	99.6%	100%	100%	99.7%	?Long QT syndrome 11, 611820
AKR1C1	100%	100%	100%	100%	99.5%	
AKR1C2	96.7%	92.5%	99.9%	99.7%	98.6%	46XY sex reversal 8, 614279
AKR1D1	88.4%	88.4%	100%	100%	99.8%	Bile acid synthesis defect, congenital, 2, 235555

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AKT1	100%	100%	100%	100%	99.1%	Proteus syndrome, somatic mosaic, 176920;Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Ovarian cancer, somatic, 167000
AKT2	98.3%	95%	100%	100%	99.4%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
AKT3	94.6%	94.6%	100%	100%	99.7%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100%	100%	100%	100%	99.4%	Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740
ALAS2	100%	100%	99%	90.9%	70.9%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752
ALB	100%	99.1%	100%	100%	99.9%	?[Dysalbuminemic hypertriiodothyroninemia], 615999;Analbuminemia, 616000;[Dysalbuminemic hyperthyroxinemia], 615999
ALDH18A1	100%	100%	100%	100%	99.6%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ALDH1A2	88.9%	88.9%	100%	100%	99.5%	Diaphragmatic hernia 4, with cardiovascular defects, 620025
ALDH1A3	94.4%	94.4%	100%	99.9%	99.4%	Microphthalmia, isolated 8, 615113
ALDH1B1	100%	100%	100%	100%	99.5%	
ALDH2	100%	99.9%	100%	100%	99.4%	{Sublingual nitroglycerin, susceptibility to poor response to}, 610251;Alcohol sensitivity, acute, 610251;{Hangover, susceptibility to}, 610251
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200
ALDH4A1	97.9%	96.1%	100%	100%	99.5%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	100%	99.5%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH6A1	95.2%	95.2%	100%	100%	99.7%	Methylmalonate semialdehyde dehydrogenase deficiency, 614105
ALDH7A1	97.2%	97.2%	100%	100%	99.8%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALDOA	100%	100%	100%	100%	99.4%	Glycogen storage disease XII, 611881
ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
ALG1	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ik, 608540
ALG10	100%	100%	100%	100%	99.8%	
ALG11	91%	91%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ip, 613661

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ALG12	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	100%	99.2%	90.7%	72.3%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110
ALG5	91.6%	91.6%	100%	100%	99.9%	Polycystic kidney disease 7, 620056
ALG6	100%	100%	100%	100%	99.9%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	78.4%	77.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	99.9%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ALK	100%	100%	100%	100%	99.4%	{Neuroblastoma, susceptibility to, 3}, 613014
ALKBH1	90.6%	90.6%	100%	100%	99.8%	
ALKBH8	96.3%	96.3%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	100%	100%	99.8%	Alstrom syndrome, 203800
ALOX12B	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100%	100%	100%	100%	99.4%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPI	100%	100%	100%	99.9%	99.3%	
ALPK1	100%	100%	100%	100%	99.7%	ROSAH syndrome, 614979
ALPK3	100%	100%	100%	100%	99.5%	Cardiomyopathy, familial hypertrophic 27, 618052
ALPL	100%	100%	100%	100%	99.3%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALS2	97%	96.3%	100%	100%	99.8%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100
ALX1	100%	100%	100%	100%	99.7%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	100%	99.5%	98.7%	Frontonasal dysplasia 1, 136760

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ALX4	100%	100%	100%	100%	99.4%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMACR	100%	100%	100%	99.8%	99.4%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMBN	100%	100%	100%	100%	99.9%	Amelogenesis imperfecta, type IF, 616270
AMELX	100%	100%	98.3%	89.3%	72.6%	Amelogenesis imperfecta, type 1E, 301200
AMER1	100%	100%	98.9%	90.5%	71.9%	Osteopathia striata with cranial sclerosis, 300373
AMFR	100%	100%	100%	100%	99.6%	Spastic paraplegia 89, autosomal recessive, 620379
AMH	100%	100%	100%	99.9%	99%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	100%	100%	100%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AMMECR1	99.9%	97.7%	99.1%	88.8%	71.2%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMN	100%	100%	100%	99.9%	99.1%	Imerslund-Grasbeck syndrome 2, 618882
AMOTL1	100%	100%	100%	100%	99.5%	Craniofaciocardiohepatic syndrome, 621192
AMPD1	100%	100%	100%	100%	99.7%	Myopathy due to myoadenylate deaminase deficiency, 615511

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AMPD2	100%	100%	100%	99.9%	99.6%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
AMPD3	100%	100%	100%	100%	99.8%	[AMP deaminase deficiency, erythrocytic], 612874
AMT	100%	100%	100%	99.9%	99.3%	Glycine encephalopathy 2, 620398
AMTN	100%	100%	100%	100%	99.8%	?Amelogenesis imperfecta, type IIIB, 617607
ANAPC1	100%	100%	100%	99.9%	99.5%	Rothmund-Thomson syndrome, type 1, 618625
ANAPC7	91%	91%	100%	100%	99.6%	Ferguson-Bonni neurodevelopmental syndrome, 619699
ANG	100%	100%	100%	100%	99.1%	Amyotrophic lateral sclerosis 9, 611895
ANGPT1	100%	100%	100%	100%	99.6%	?Angioedema, hereditary, 5, 619361
ANGPT2	100%	100%	100%	100%	99.7%	Lymphatic malformation 10, 619369
ANGPTL3	100%	100%	100%	100%	99.6%	Hypobetalipoproteinemia, familial, 2, 605019
ANGPTL4	100%	100%	100%	100%	99.4%	Plasma triglyceride level QTL, low, 615881
ANK1	100%	100%	100%	100%	99.5%	Spherocytosis, type 1, 182900
ANK2	100%	100%	100%	100%	99.7%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	99.7%	99.7%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 37, 615493

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ANKFY1	100%	100%	100%	100%	99.5%	
ANKH	100%	100%	100%	100%	99.4%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKLE2	100%	100%	100%	99.8%	99.1%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD1	100%	100%	100%	99.9%	99%	
ANKRD11	100%	100%	100%	100%	99.2%	KBG syndrome, 148050
ANKRD17	100%	100%	100%	100%	99.6%	Chopra-Amiel-Gordon syndrome, 619504
ANKRD26	100%	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000
ANKRD31	100%	100%	100%	99.9%	99.8%	
ANKS1B	100%	100%	100%	100%	99.8%	
ANKS6	100%	99.9%	100%	100%	99.5%	Nephronophthisis 16, 615382
ANLN	98.2%	98.2%	100%	100%	99.7%	Focal segmental glomerulosclerosis 8, 616032
ANO1	100%	100%	100%	99.9%	98.7%	Moyamoya disease 7, 620687;?Intestinal dysmotility syndrome, 620045
ANO10	94.9%	94.9%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	100%	100%	100%	100%	99.8%	Dystonia 24, 615034
ANO4	100%	100%	100%	100%	99.7%	
ANO5	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307;Miyoshi muscular dystrophy 3, 613319;Gnathodiaphyseal dysplasia, 166260
ANO6	95.9%	94.9%	100%	100%	99.8%	Scott syndrome, 262890

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ANOS1	100%	100%	98.7%	89.8%	71.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	100%	100%	100%	99.9%	99.4%	GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	94.3%	94.3%	100%	100%	99.8%	Hyaline fibromatosis syndrome, 228600
ANXA11	100%	98.9%	100%	100%	99.5%	Amyotrophic lateral sclerosis 23, 617839;Inclusion body myopathy and brain white matter abnormalities, 619733
AOPEP	100%	100%	100%	100%	99.5%	Dystonia 31, 619565
AP1B1	93.7%	91.9%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1G1	100%	100%	100%	100%	99.7%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100%	100%	100%	100%	99.2%	MEDNIK syndrome, 609313
AP1S2	100%	100%	99.3%	92.3%	76.1%	Pettigrew syndrome, 304340
AP1S3	90.6%	90.6%	100%	100%	99.9%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP2M1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 60, with seizures, 618587

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AP2S1	82.5%	82.5%	100%	99.9%	98.8%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100%	100%	100%	100%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100%	100%	100%	100%	99.8%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100%	100%	100%	100%	99.9%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	100%	100%	99.7%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	79.1%	79.1%	100%	99.9%	99.4%	Spastic paraplegia 52, autosomal recessive, 614067
AP5B1	100%	100%	100%	100%	99.4%	
AP5M1	100%	100%	100%	100%	99.9%	
AP5Z1	91.2%	91%	100%	100%	99.5%	Spastic paraplegia 48, autosomal recessive, 613647

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

APC	100%	100%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;Brain tumor-polyposis syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100
APC2	100%	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
APCDD1	100%	100%	100%	100%	99.5%	Hypotrichosis 1, 605389
APOA1	100%	100%	100%	99.8%	99.3%	Hypoalphalipoproteinemia, primary, 2, 618463;Amyloidosis, hereditary systemic 3, 620657;Hypoalphalipoproteinemia, primary, 2, intermediate, 619836
APOA2	100%	100%	100%	100%	99.8%	[?Apolipoprotein A-II deficiency], 621417;{Hypercholesterolemia, familial, modifier of}, 143890
APOA4	100%	100%	100%	99.9%	98.8%	Tubulointerstitial kidney disease, autosomal dominant 6, 621106
APOA5	100%	100%	100%	100%	99.4%	Hyperchylomicronemia, late-onset, 144650;{Hypertriglyceridemia, susceptibility to}, 145750

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

APOB	100%	100%	100%	100%	99.7%	Hypercholesterolemia, familial, 2, 144010;Hypobetalipoproteinemia, 615558
APOC2	100%	100%	100%	100%	98.3%	Hyperlipoproteinemia, type Ib, 207750
APOC3	100%	100%	100%	100%	99.9%	Apolipoprotein C-III deficiency, 614028
APOE	93.8%	93.4%	100%	100%	99.1%	Alzheimer disease 2, 104310;Sea-blue histiocyte disease, 269600;{?Alzheimer disease, protection against, due to APOE3-Christchurch}, 607822;{Coronary artery disease, severe, susceptibility to}, 617347;Lipoprotein glomerulopathy, 611771;{?Macular degeneration, age-related}, 603075;Hyperlipoproteinemia, type III, 617347
APOL1	100%	100%	100%	100%	99.5%	{Glomerulosclerosis, focal segmental, 4, susceptibility to}, 612551
APOLD1	100%	99.4%	100%	99.9%	97.5%	?Bleeding disorder, vascular-type, 620715
APOO	80.2%	80.1%	98.6%	91.1%	74%	
APP	100%	100%	100%	100%	99.8%	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714;Alzheimer disease 1, familial, 104300
APRT	100%	100%	100%	100%	99.1%	Adenine phosphoribosyltransferase deficiency, 614723
APTX	100%	100%	100%	100%	99.7%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AQP2	100%	100%	100%	100%	99.8%	Diabetes insipidus, nephrogenic, 2, 125800
AQP4	100%	100%	100%	99.9%	99.8%	?Megalencephalic leukoencephalopathy with subcortical cysts 4, remitting, 620448
AQP5	100%	100%	100%	100%	99.5%	Palmoplantar keratoderma, Bothnian type, 600231
AR	100%	100%	98.4%	87.6%	66.9%	Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;{Prostate cancer, susceptibility to}, 301120;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633
ARAF	100%	99.9%	98.7%	88.4%	67.6%	
ARCN1	96.3%	96.1%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ARF1	100%	100%	100%	100%	99.8%	Periventricular nodular heterotopia 8, 618185
ARF3	100%	100%	100%	100%	99.1%	
ARFGEF1	100%	100%	100%	100%	99.9%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964
ARFGEF2	100%	100%	100%	100%	99.5%	Periventricular heterotopia with microcephaly, 608097
ARG1	93%	93%	100%	100%	99.8%	Argininemia, 207800
ARHGAP19	94.5%	94.1%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2KK, 621466
ARHGAP24	100%	100%	100%	99.9%	99.2%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ARHGAP26	100%	100%	100%	100%	99.6%	Leukemia, juvenile myelomonocytic, somatic, 607785
ARHGAP29	100%	100%	100%	100%	99.8%	
ARHGAP31	100%	100%	100%	100%	99.6%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100%	100%	100%	100%	99.5%	
ARHGDIA	100%	100%	100%	100%	99%	Nephrotic syndrome, type 8, 615244
ARHGEF1	100%	99.8%	100%	99.9%	99%	?Immunodeficiency 62, 618459
ARHGEF10	100%	100%	100%	100%	99.7%	?Slowed nerve conduction velocity, AD, 608236
ARHGEF15	100%	100%	100%	100%	99.2%	Brain small vessel disease 5 with osteoporosis, 621331
ARHGEF18	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 78, 617433
ARHGEF2	97.9%	97.9%	100%	100%	99.3%	?Neurodevelopmental disorder with midbrain and hindbrain malformations, 617523
ARHGEF28	100%	100%	100%	100%	99.7%	
ARHGEF6	93.6%	93.6%	99.2%	90.8%	73.8%	
ARHGEF9	96.9%	96.9%	99.3%	90.1%	70.9%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100%	100%	100%	99.9%	98.9%	Coffin-Siris syndrome 2, 614607
ARID1B	98.7%	98.4%	100%	99.5%	96.7%	Coffin-Siris syndrome 1, 135900
ARID2	100%	100%	100%	100%	99.7%	Coffin-Siris syndrome 6, 617808
ARIH1	100%	100%	100%	100%	99.8%	
ARL13B	93.4%	93.3%	100%	100%	99.7%	Joubert syndrome 8, 612291

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ARL2	100%	100%	100%	100%	99%	?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 1, 619082
ARL2BP	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 82 with or without situs inversus, 615434
ARL3	86.8%	86.8%	100%	100%	99.8%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161
ARL6	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARL6IP1	100%	100%	100%	100%	99.9%	Spastic paraplegia 61, autosomal recessive, 615685
ARMC12	100%	100%	100%	99.8%	99.5%	Spermatogenic failure 90, 620744
ARMC2	100%	100%	100%	100%	99.8%	Spermatogenic failure 38, 618433
ARMC5	100%	100%	100%	100%	99.4%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
ARMC9	96.3%	94.2%	100%	100%	99.3%	Joubert syndrome 30, 617622
ARNT2	91%	91%	100%	100%	99.5%	?Webb-Dattani syndrome, 615926
ARPC1B	99.4%	95.9%	100%	100%	99.3%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
ARPC4	100%	100%	100%	100%	99.9%	Developmental delay, language impairment, and ocular abnormalities, 620141

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ARPC5	100%	100%	100%	100%	100%	Immunodeficiency 113 with autoimmunity and autoinflammation, 620565
ARR3	100%	100%	99.5%	90.6%	69.9%	Myopia 26, X-linked, female-limited, 301010
ARSA	100%	100%	100%	99.9%	99%	Metachromatic leukodystrophy, 250100
ARSB	100%	100%	100%	100%	99.3%	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200
ARSG	100%	100%	100%	100%	99.1%	Usher syndrome, type IV, 618144
ARSK	100%	100%	100%	100%	99.8%	Mucopolysaccharidosis , type X, 619698
ARSL	100%	100%	98.7%	90.2%	71.8%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	87.3%	86.5%	100%	100%	99.8%	Developmental and epileptic encephalopathy 38, 617020
ARX	99.9%	98.4%	96.3%	80.9%	59.5%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100%	100%	100%	99.9%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ASB10	100%	100%	100%	100%	99.7%	Glaucoma 1, open angle, F, 603383
ASCC1	86.6%	86.6%	100%	100%	99.6%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266
ASCC3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 81, 620700
ASCL1	100%	100%	100%	99.9%	97.6%	
ASH1L	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASIP	100%	100%	100%	100%	99.9%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742;[Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	100%	100%	100%	100%	99.1%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	100%	99.8%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	100%	100%	99.8%	Canavan disease, 271900
ASPH	100%	100%	100%	100%	99.7%	Traboulsi syndrome, 601552
ASPM	97.9%	97.9%	100%	100%	99.7%	Microcephaly 5, primary, autosomal recessive, 608716
ASPRV1	100%	100%	100%	100%	99.2%	Ichthyosis, lamellar, autosomal dominant, 146750
ASPSCR1	100%	100%	100%	100%	99.2%	Alveolar soft-part sarcoma, 606243
ASRGL1	100%	100%	100%	100%	100%	
ASS1	92.3%	90%	100%	100%	99.6%	Citrullinemia, 215700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ASTL	100%	100%	100%	100%	99.7%	?Oocyte/zygote/embryo maturation arrest 11, 619643
ASTN1	96.6%	96.6%	100%	100%	99.5%	
ASXL1	100%	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL2	100%	100%	100%	100%	99.6%	Shashi-Pena syndrome, 617190
ASXL3	100%	100%	100%	100%	99.6%	Bainbridge-Ropers syndrome, 615485
ATAD1	96.8%	96.8%	100%	100%	100%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	100%	99.9%	98.7%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATAD3B	94.5%	94.5%	99.9%	98.7%	96.3%	
ATCAY	100%	100%	100%	100%	99.2%	Ataxia, cerebellar, Cayman type, 601238
ATF3	100%	100%	100%	100%	99.6%	
ATF6	90.9%	90.9%	100%	100%	99.7%	Achromatopsia 7, 616517
ATG4A	100%	99.7%	99.7%	91.9%	75%	
ATG4D	100%	100%	100%	100%	99.2%	Spermatogenic failure 101, 621269
ATG5	90.4%	90.4%	100%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 25, 617584
ATG7	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100%	100%	100%	100%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATL1	100%	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATL3	100%	100%	100%	100%	99.7%	Neuropathy, hereditary sensory, type IF, 615632
ATM	100%	100%	100%	100%	99.7%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
ATN1	98.2%	98.2%	100%	100%	98.7%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	100%	100%	100%	100%	99.6%	?Deafness, autosomal dominant 89, 620284
ATOH7	100%	100%	100%	100%	99.6%	Persistent hyperplastic primary vitreous, autosomal recessive, 221900
ATP11A	100%	100%	100%	100%	99.7%	?Auditory neuropathy, autosomal dominant 2, 620384;?Leukodystrophy, hypomyelinating, 24, 619851;Deafness, autosomal dominant 84, 619810
ATP11C	100%	100%	99.2%	92.9%	76.1%	?Hemolytic anemia, congenital, X-linked, 301015
ATP13A2	100%	100%	100%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATP13A3	97%	97%	100%	100%	99.8%	Pulmonary hypertension, primary, 5, 265400
ATP1A1	97.2%	95.4%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481
ATP1A3	100%	100%	100%	100%	99.2%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2A1	100%	100%	100%	100%	99.3%	Brody myopathy, 601003
ATP2A2	100%	100%	100%	100%	99.6%	Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236
ATP2B1	100%	100%	100%	99.9%	99.6%	Intellectual developmental disorder, autosomal dominant 66, 619910

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATP2B2	100%	100%	100%	100%	99.2%	Deafness, autosomal dominant 82, 619804
ATP2B3	100%	100%	98.7%	88.2%	69%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP2C1	96.2%	96.2%	100%	100%	99.8%	Hailey-Hailey disease, 169600
ATP5F1A	100%	100%	100%	99.9%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4A, 620358;?Combined oxidative phosphorylation deficiency 22, 616045;?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4B, encephalopathic type, 615228
ATP5F1B	96.8%	96.6%	100%	100%	99.7%	?Hypermetabolism due to uncoupled mitochondrial oxidative phosphorylation 2, 620085;{Dystonia 38, susceptibility to}, 621502
ATP5F1C	100%	99.7%	100%	100%	99.6%	
ATP5F1D	100%	100%	100%	99.9%	98.9%	Mitochondrial complex V (ATP synthase) deficiency, 618120
ATP5F1E	100%	100%	100%	100%	100%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053
ATP5IF1	89.1%	89.1%	100%	100%	99.4%	
ATP5MC1	86.7%	82.6%	100%	100%	98.8%	
ATP5MC2	100%	100%	100%	100%	99.8%	
ATP5MC3	100%	100%	100%	100%	99.8%	Dystonia, early-onset, and/or spastic paraplegia, 619681
ATP5ME	100%	100%	100%	100%	99%	
ATP5MF	100%	100%	100%	100%	100%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATP5MG	87.9%	87.9%	100%	100%	99.6%	
ATP5MGL	100%	100%	100%	100%	98.3%	
ATP5MK	100%	100%	100%	100%	99.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 6, 618683
ATP5PB	100%	100%	100%	100%	99.7%	
ATP5PD	100%	100%	100%	100%	100%	
ATP5PF	100%	100%	100%	100%	99.9%	
ATP5PO	100%	100%	100%	100%	99.7%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359
ATP6AP1	99.6%	97.5%	98.9%	89.5%	70.5%	Immunodeficiency 47, 300972
ATP6AP2	100%	100%	99.3%	91.2%	74.9%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0A2	100%	100%	100%	100%	99.6%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0A4	100%	100%	100%	99.9%	99%	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATP6V0C	100%	100%	100%	100%	99.8%	Epilepsy, early-onset, 3, with or without developmental delay, 620465
ATP6V1A	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012
ATP6V1B1	100%	100%	100%	100%	99.5%	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300
ATP6V1B2	94.3%	94.3%	100%	100%	99.8%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP6V1C1	100%	100%	100%	100%	100%	
ATP6V1E1	100%	100%	100%	100%	99.6%	Cutis laxa, autosomal recessive, type IIC, 617402
ATP7A	95%	94.8%	99.4%	92.2%	74.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
ATP8A2	100%	100%	100%	100%	99.6%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATP8B1	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic 1, 211600;Cholestasis, intrahepatic, of pregnancy, 1, 147480;Cholestasis, benign recurrent intrahepatic, 243300
ATP9A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242
ATPAF1	100%	100%	100%	99.9%	98.1%	
ATPAF2	100%	100%	100%	100%	99.5%	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273
ATR	100%	100%	99.9%	99.7%	99.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	100%	100%	99.1%	91.6%	74.3%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
ATXN1	100%	100%	100%	99.9%	99.4%	Spinocerebellar ataxia 1, 164400
ATXN10	100%	100%	100%	100%	99.4%	Spinocerebellar ataxia 10, 603516

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ATXN2	100%	100%	100%	99.9%	99.4%	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090;Spinocerebellar ataxia 2, 183090;{Parkinson disease, late-onset, susceptibility to}, 168600
ATXN2L	100%	100%	100%	99.8%	99.2%	
ATXN3	97%	97%	100%	99.9%	99.6%	{Parkinson disease, late-onset, susceptibility to}, 168600;Machado-Joseph disease, 109150
ATXN7	100%	100%	100%	100%	99.1%	Spinocerebellar ataxia 7, 164500
ATXN7L3	100%	100%	100%	100%	99.6%	Harel-Tora neurodevelopmental syndrome, 621377
ATXN8OS						{Parkinson disease, susceptibility to}, 168600;Spinocerebellar ataxia 8, 608768
AUH	100%	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type I, 250950
AURKC	100%	100%	100%	100%	99.5%	Spermatogenic failure 5, 243060
AUTS2	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVIL	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 21, 618594
AVP	100%	100%	100%	100%	99.8%	Diabetes insipidus, neurohypophyseal, 125700
AVPR2	100%	100%	98.8%	89%	71.9%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

AXIN1	100%	100%	100%	100%	99.5%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
AXIN2	100%	100%	100%	100%	99.2%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
AXL	100%	100%	100%	100%	99.1%	
B2M	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 6, 620659;Immunodeficiency 43, 241600
B3GALNT1	100%	100%	100%	100%	100%	[Blood group, P1PK system, P(k) phenotype], 111400;[Blood group, globoside system], 615021
B3GALNT2	92.6%	92.6%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181
B3GALT6	100%	98.6%	100%	99.9%	98.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B3GAT3	94.7%	93.8%	100%	100%	99.8%	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

B3GLCT	100%	100%	100%	100%	99.9%	Peters-plus syndrome, 261540
B4GALNT1	100%	100%	100%	100%	99.5%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALNT2	100%	100%	100%	99.9%	99.3%	[Blood group, Sid system], 615018;Sd(a) polyagglutination syndrome, 615018
B4GALT1	91%	88.3%	100%	100%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	86.8%	86.8%	100%	100%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	99.7%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BAAT	100%	100%	100%	100%	99.8%	Hypercholanemia, familial 3, 619232
BACH2	100%	100%	100%	100%	99.4%	Immunodeficiency 60 and autoimmunity, 618394

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BAG3	100%	100%	100%	100%	99.5%	?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095
BAG5	100%	100%	100%	100%	98.8%	Cardiomyopathy, dilated, 2F, 619747
BAIAP2	100%	100%	100%	99.9%	99.3%	Developmental and epileptic encephalopathy 120, 621468
BAIAP3	100%	100%	100%	99.9%	99.4%	
BANF1	100%	100%	100%	99.8%	98.5%	Nestor-Guillermo progeria syndrome, 614008
BAP1	100%	100%	100%	100%	99.5%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BARD1	100%	100%	100%	100%	99.7%	{Breast cancer, susceptibility to}, 114480
BAX	100%	100%	100%	100%	98.4%	Colorectal cancer, somatic, 114500;T-cell acute lymphoblastic leukemia, somatic, 613065
BAZ2B	100%	100%	100%	99.9%	99.3%	
BBIP1	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 18, 615995
BBS1	93.7%	93.7%	100%	100%	99.3%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 10, 615987

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.8%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCAP31	91.5%	86.7%	98.7%	88.7%	70.4%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100%	100%	100%	100%	99.8%	Hengel-Maroofian-Schols syndrome, 619641
BCAT1	97%	97%	100%	100%	99.7%	
BCAT2	100%	100%	100%	100%	99.4%	Hypervalinemia and hyperleucine-isoleucinemia, 618850
BCHE	95.9%	95.9%	100%	100%	99.9%	Butyrylcholinesterase deficiency, 617936;{Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936
BCKDHA	86.3%	86.3%	100%	99.9%	99.3%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89.9%	87.7%	100%	99.9%	99.3%	Maple syrup urine disease, type Ib, 620698
BCKDK	100%	100%	100%	100%	99.6%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BCL10	100%	100%	100%	100%	99.7%	{Lymphoma, follicular, somatic}, 605027;?Immunodeficiency 37, 616098;{Sezary syndrome, somatic};{Male germ cell tumor, somatic}, 273300;Lymphoma, MALT, somatic, 137245;{Mesothelioma, somatic}, 156240
BCL11A	100%	100%	100%	99.9%	98.9%	Dias-Logan syndrome, 617101
BCL11B	100%	99.8%	100%	99.9%	98%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCL2	92.5%	92.5%	100%	99.9%	99.2%	
BCL7A	100%	100%	100%	99.9%	98.8%	
BCL9L	100%	100%	100%	100%	99.3%	
BCO1	100%	100%	100%	100%	99.6%	?Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300
BCOR	100%	100%	98.9%	89.4%	69.8%	Microphthalmia, syndromic 2, 300166
BCORL1	100%	100%	98.5%	87.1%	67.3%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BDP1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 112, 618257
BEAN1	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia 31, 117210

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BEST1	100%	100%	100%	99.9%	98.6%	Macular dystrophy, vitelliform, 2, 153700;Retinitis pigmentosa 50, 613194;?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220;Retinitis pigmentosa, concentric, 613194;Vitreoretinochoroidopathy, 193220;Bestrophinopathy, autosomal recessive, 611809
BET1	100%	100%	100%	100%	99.8%	Muscular dystrophy, congenital, with rapid progression, 254100
BFSP1	100%	100%	100%	99.9%	99.4%	Cataract 33, multiple types, 611391
BFSP2	100%	100%	100%	99.9%	99.1%	Cataract 12, multiple types, 611597
BGN	99.9%	98.4%	97.8%	87.7%	69.5%	Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106
BHLHA9	100%	100%	100%	100%	99.4%	?Camptosynpolydactyly, complex, 607539;Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432
BHLHE22	100%	100%	100%	99.8%	97.3%	
BICC1	100%	100%	100%	100%	99.8%	{Renal dysplasia, cystic, susceptibility to}, 601331
BICD1	100%	100%	100%	100%	99.4%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BICD2	100%	100%	100%	100%	99.4%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BICRA	100%	100%	100%	99.8%	98.4%	Coffin-Siris syndrome 12, 619325
BIN1	100%	100%	100%	100%	99.3%	Centronuclear myopathy 2, 255200
BLK	100%	100%	100%	100%	99.4%	Maturity-onset diabetes of the young, type 11, 613375
BLM	94.7%	94.6%	100%	100%	99.9%	Bloom syndrome, 210900
BLNK	94.4%	94.4%	100%	100%	99.8%	?Agammaglobulinemia 4, 613502
BLOC1S1	100%	100%	100%	100%	99.1%	
BLOC1S3	100%	100%	100%	100%	98.5%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 9, 614171
BLTP1	100%	100%	100%	100%	99.8%	Alkuraya-Kucinskas syndrome, 617822
BLVRA	95.2%	95.2%	100%	100%	99.8%	Hyperbiliverdinemia, 614156
BMP1	100%	100%	100%	100%	99.3%	Osteogenesis imperfecta, type XIII, 614856
BMP10	100%	100%	100%	100%	99.9%	
BMP15	100%	100%	98.6%	90.2%	70.4%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BMP2	100%	100%	100%	99.8%	97.7%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BMP4	100%	100%	100%	100%	99.5%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BMP5	100%	100%	100%	100%	99.5%	
BMP6	100%	100%	100%	99.8%	98.9%	{Iron overload, susceptibility to}, 620121
BMP7	100%	100%	100%	100%	99%	
BMPER	100%	100%	100%	100%	99.6%	Diaphanospondylodysostosis, 608022
BMPR1A	96.5%	96.5%	100%	100%	99.8%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900
BMPR1B	95.6%	94.1%	100%	100%	99.7%	Acromesomelic dysplasia 3, 609441;Brachydactyly, type A2, 112600;Brachydactyly, type A1, D, 616849
BMPR2	100%	100%	100%	100%	99.7%	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600;Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600;Pulmonary venoocclusive disease 1, 265450
BMS1	100%	100%	100%	100%	99.8%	?Aplasia cutis congenita, nonsyndromic, 107600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BNC1	100%	100%	100%	100%	99.8%	?Premature ovarian failure 16, 618723
BNC2	96.6%	96.6%	100%	100%	99.8%	Lower urinary tract obstruction, congenital, 618612
BNIP1	100%	100%	100%	100%	99.9%	Spondylopeiphyseal dysplasia, Holling type, 621345
BOLA1	100%	100%	100%	100%	99.2%	
BOLA2	100%	100%	100%	100%	100%	
BOLA3	100%	100%	100%	100%	98.9%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BORCS5	89.3%	89.3%	100%	100%	99.5%	
BORCS8	84.1%	84.1%	100%	99.7%	98.8%	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987
BPGM	100%	100%	100%	100%	99.9%	Erythrocytosis, familial, 8, 222800
BPNT2	94.9%	94.9%	100%	100%	99.5%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
BPTF	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755;{Kaposi sarcoma, susceptibility to}, 148000
BPY2	50%	50%	45.9%	37.5%	24.4%	
BPY2B	50%	49.8%	45.9%	35.9%	21.2%	
BPY2C	50%	50%	44.7%	36.2%	24.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BRAF	100%	100%	100%	99.8%	98.2%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
BRAT1	97.3%	96.5%	100%	99.9%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRCA1	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070
BRD4	100%	100%	100%	99.9%	98.7%	Cornelia de Lange syndrome 6, 620568

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BRDT	100%	100%	100%	100%	99.9%	?Spermatogenic failure 21, 617644
BRF1	100%	100%	100%	100%	99.5%	Cerebellofaciodental syndrome, 616202
BRIP1	96%	96%	100%	100%	99.8%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
BRPF1	100%	100%	100%	100%	99.6%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	100%	100%	100%	99.9%	99.1%	
BRWD1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 51, 620438
BRWD3	100%	100%	99.3%	91.7%	73.7%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	100%	100%	100%	100%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BSN	100%	99.9%	100%	99.9%	99.3%	
BSND	100%	100%	100%	100%	99.8%	Sensorineural deafness with mild renal dysfunction, 602522;Bartter syndrome, type 4a, 602522
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

BTG4	100%	100%	100%	99.9%	99.8%	Oocyte/zygote/embryo maturation arrest 8, 619009
BTK	100%	99.4%	98.9%	90.7%	72.8%	Agammaglobulinemia, X-linked 1, 300755;Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200
BTRC	95.7%	92.5%	100%	100%	99.5%	
BUB1	97.3%	97.3%	100%	100%	99.7%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183
BUB1B	96.3%	96.3%	100%	100%	99.8%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
BUB3	100%	100%	100%	100%	99.6%	
BUD13	100%	100%	100%	100%	99.4%	Achalasia-progeroid syndrome, 621123
C10orf71	100%	100%	100%	99.9%	99.3%	Cardiomyopathy, dilated, 1QQ, 621251
C12orf57	100%	100%	100%	99.8%	98.3%	Temtamy syndrome, 218340
C14orf39	100%	100%	100%	100%	99.7%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203
C18orf32	100%	100%	100%	100%	99%	?Glycosylphosphatidylinositol biosynthesis defect 25, 619985

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
C19orf44	100%	100%	100%	100%	99.4%	
C1GALT1C1	100%	100%	99.4%	93.2%	73.1%	Hemolytic uremic syndrome, atypical, 8, with rhizomelic short stature, 301110;Tn polyagglutination syndrome, somatic, 300622
C1QA	76.9%	73.9%	100%	100%	99.6%	C1q deficiency 1, 613652
C1QB	77.2%	77.2%	100%	100%	99.3%	C1q deficiency 2, 620321
C1QBP	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 33, 617713
C1QC	99.7%	98%	100%	100%	98.7%	C1q deficiency 3, 620322
C1QTNF5	100%	100%	100%	100%	99.6%	Retinal degeneration, late-onset, autosomal dominant, 605670
C1R	100%	98.1%	100%	99.9%	99.5%	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	100%	100%	100%	100%	99.5%	C1s deficiency, 613783;Ehlers-Danlos syndrome, periodontal type, 2, 617174
C2	100%	100%	100%	100%	99.5%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489
C2CD3	96%	96%	100%	100%	99.5%	Orofaciodigital syndrome XIV, 615948
C2CD6	100%	100%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

C2orf69	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 53, 619423
C3	97%	97%	100%	99.9%	99.2%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C3orf52	96.1%	96.1%	100%	100%	99.9%	Hypotrichosis 15, 620177
C4A	99.5%	99.1%	99.2%	97.9%	94.6%	[Blood group, Rodgers], 614374;C4a deficiency, 614380
C4B	100%	99.9%	99.2%	97.8%	94.3%	C4B deficiency, 614379
C5	100%	100%	100%	100%	99.8%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
C6	100%	100%	100%	100%	99.9%	C6 deficiency, 612446
C7	86.4%	85.8%	100%	100%	99.6%	C7 deficiency, 610102
C8A	100%	100%	100%	100%	99.9%	C8 deficiency, type I, 613790
C8B	100%	100%	100%	100%	99.8%	C8 deficiency, type II, 613789
C8G	100%	100%	100%	100%	99.4%	
C9	99.3%	99.3%	100%	99.9%	99.6%	C9 deficiency, 613825;{Macular degeneration, age-related, 15, susceptibility to}, 615591
C9orf72	100%	100%	100%	100%	99.9%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550
CA12	100%	100%	100%	100%	99.5%	Hyperchlorhidrosis, isolated, 143860

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CA2	100%	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA4	84.7%	80.6%	100%	100%	99.6%	
CA5A	84.7%	84.7%	100%	100%	99.6%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 34, 613227
CABIN1	100%	100%	100%	100%	99.5%	
CABP2	100%	100%	100%	99.7%	97.3%	Deafness, autosomal recessive 93, 614899
CABP4	100%	100%	100%	100%	99.5%	Cone-rod synaptic disorder, congenital nonprogressive, 610427
CACHD1	100%	100%	100%	100%	99.7%	Davis-Wells syndrome, 621566
CACNA1A	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500
CACNA1B	97.2%	97.2%	100%	99.9%	99%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CACNA1C	100%	100%	100%	100%	99.5%	Timothy syndrome, 601005;Long QT syndrome 8, 618447;Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029;Brugada syndrome 3, 611875
CACNA1D	100%	100%	100%	100%	99.7%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 69, 618285
CACNA1F	100%	100%	98%	86.9%	67.2%	Cone-rod dystrophy, X-linked, 3, 300476;Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071;Aland Island eye disease, 300600
CACNA1G	100%	100%	100%	99.9%	99.4%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1H	99.6%	99%	100%	99.9%	99%	{Epilepsy, childhood absence, susceptibility to, 6}, 611942;Hyperaldosteronism, familial, type IV, 617027;{Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CACNA1I	100%	100%	100%	99.8%	98.5%	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114
CACNA1S	100%	100%	100%	100%	99.4%	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580;Congenital myopathy 18 due to dihydropyridine receptor defect, 620246;Hypokalemic periodic paralysis, type 1, 170400;{Malignant hyperthermia susceptibility 5}, 601887
CACNA2D1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 110, 620149
CACNA2D2	100%	100%	100%	100%	99.3%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNA2D4	100%	100%	100%	100%	99.5%	Retinal cone dystrophy 4, 610478
CACNB2	100%	100%	100%	100%	99.3%	Brugada syndrome 4, 611876
CACNB4	100%	100%	100%	100%	99.8%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682;?Episodic ataxia, type 5, 613855;{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682
CACNG2	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal dominant 10, 614256
CAD	99.7%	98.5%	100%	100%	99.4%	Developmental and epileptic encephalopathy 50, 616457

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CADM3	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2FF, 619519
CALCRL	100%	100%	100%	100%	99.8%	?Lymphatic malformation 8, 618773
CALM1	100%	100%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916;Long QT syndrome 14, 616247
CALM2	73.5%	73.5%	100%	99.9%	98.9%	Long QT syndrome 15, 616249
CALM3	100%	100%	100%	100%	99.7%	Long QT syndrome 16, 618782;?Ventricular tachycardia, catecholaminergic polymorphic 6, 618782
CALR	100%	100%	100%	100%	99.4%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CALR3	100%	100%	100%	100%	99.4%	
CAMK2A	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095
CAMK2B	100%	100%	100%	99.9%	99%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2D	97.2%	97.2%	100%	100%	99.6%	
CAMK2G	99.7%	98.1%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	100%	100%	100%	100%	99.7%	
CAMLG	93.3%	93.3%	100%	100%	99.8%	?Congenital disorder of glycosylation, type IIz, 620201

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CAMSAP1	100%	100%	100%	99.9%	99.3%	Cortical dysplasia, complex, with other brain malformations 12, 620316
CAMTA1	100%	100%	100%	100%	99.2%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	100%	100%	100%	100%	99.7%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CAP2	82%	82%	100%	99.9%	99.8%	Cardiomyopathy, dilated, 2I, 620462
CAPN1	100%	100%	100%	100%	99.2%	Spastic paraplegia 76, autosomal recessive, 616907
CAPN10	100%	100%	100%	99.9%	99.4%	{Diabetes mellitus, noninsulin-dependent 1}, 601283
CAPN12	100%	100%	100%	99.9%	98.6%	
CAPN15	100%	100%	100%	99.9%	98.9%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPN3	100%	100%	100%	100%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600;Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129
CAPN5	100%	99.9%	100%	100%	99.4%	Vitreoretinopathy, neovascular inflammatory, 193235
CAPNS1	100%	100%	100%	99.9%	98.5%	Pulmonary hypertension, primary, 6, 620777

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CAPRIN1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636
CAPZA2	100%	100%	100%	100%	99.6%	
CARD10	100%	100%	100%	100%	99%	?Immunodeficiency 89 and autoimmunity, 619632
CARD11	100%	100%	100%	100%	99.5%	B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206
CARD14	100%	100%	100%	100%	99.3%	Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200
CARD8	100%	100%	100%	100%	99.7%	?Inflammatory bowel disease (Crohn disease) 30, 619079
CARD9	100%	100%	100%	100%	99.6%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100%	100%	100%	100%	99.2%	Immunodeficiency 58, 618131
CARS1	100%	100%	100%	100%	99.6%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100%	99.9%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 27, 616672

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CASK	100%	100%	99.2%	91.1%	73.2%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CASP10	100%	100%	100%	100%	99.7%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027
CASP14	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 12, 617320
CASP2	100%	99.9%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653
CASP8	97%	97%	100%	100%	99.7%	{Breast cancer, protection against}, 114480;?Caspase 8 lymphadenopathy syndrome, 607271;Hepatocellular carcinoma, somatic, 114550;{Lung cancer, protection against}, 211980
CASQ1	100%	100%	100%	100%	99.4%	Myopathy, vacuolar, with CASQ1 aggregates, 616231
CASQ2	87.5%	87.5%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CASR	100%	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CAST	100%	100%	100%	100%	99.8%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CASZ1	100%	99.6%	100%	99.9%	98.7%	
CAT	98.1%	98.1%	100%	100%	99.8%	Acatalasemia, 614097
CATIP	100%	100%	100%	100%	99.6%	?Spermatogenic failure 54, 619379
CATSPER1	100%	100%	100%	100%	98.9%	Spermatogenic failure 7, 612997
CATSPER2	100%	100%	100%	100%	99.6%	
CAV1	64.8%	64.8%	100%	100%	98.8%	Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAV3	100%	100%	100%	100%	99.2%	Myopathy, distal, Tateyama type, 614321;Creatine phosphokinase, elevated serum, 123320;Cardiomyopathy, familial hypertrophic, 192600;Rippling muscle disease 2, 606072;Long QT syndrome 9, 611818

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CAVIN1	100%	100%	100%	99.6%	97.4%	Lipodystrophy, congenital generalized, type 4, 613327
CAVIN2	100%	100%	100%	100%	99.7%	
CBFB	100%	100%	100%	100%	98.8%	Cleidocranial dysplasia 2, 620099
CBL	100%	100%	100%	100%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBLB	97.1%	97.1%	100%	100%	99.8%	Autoimmune disease, multisystem, infantile-onset, 3, 620430
CBLIF	100%	100%	100%	100%	99.8%	Intrinsic factor deficiency, 261000
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CBX1	100%	100%	100%	100%	99.9%	
CBX2	100%	100%	100%	100%	99.6%	?46XY sex reversal 5, 613080
CBY1	100%	100%	100%	100%	99.5%	
CC2D1A	100%	100%	100%	99.9%	98.9%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CCBE1	100%	100%	100%	100%	99.7%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC134	100%	100%	100%	100%	99.7%	Osteogenesis imperfecta, type XXII, 619795
CCDC141	100%	100%	100%	100%	99.8%	
CCDC146	100%	100%	100%	100%	99.8%	Spermatogenic failure 94, 620850
CCDC174	100%	100%	100%	100%	99.8%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100%	100%	100%	100%	99.7%	
CCDC22	100%	100%	98.8%	88%	69.2%	Ritscher-Schinzel syndrome 2, 300963
CCDC28B	100%	100%	100%	99.9%	98.4%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC32	100%	100%	100%	99.9%	99.5%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC34	100%	100%	100%	100%	99.8%	Spermatogenic failure 76, 620084
CCDC39	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	99.9%	99%	Ciliary dyskinesia, primary, 15, 613808
CCDC47	100%	100%	100%	100%	99.9%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC50	94.8%	94.8%	100%	100%	99.7%	?Deafness, autosomal dominant 44, 607453
CCDC62	100%	100%	100%	100%	99.6%	?Spermatogenic failure 67, 619803
CCDC65	100%	100%	100%	99.9%	99.4%	
CCDC78	100%	100%	100%	100%	99.5%	Centronuclear myopathy 4, 614807
CCDC8	99.8%	97.4%	100%	100%	99.5%	3-M syndrome 3, 614205

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CCDC82	100%	100%	100%	100%	99.8%	
CCDC88A	97.4%	97.4%	100%	100%	99.7%	PEHO syndrome-like, 617507
CCDC88C	100%	100%	100%	99.9%	98.8%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600
CCIN	100%	100%	100%	100%	99.8%	Spermatogenic failure 91, 620838
CCM2	91.1%	91.1%	100%	100%	99.3%	Cerebral cavernous malformations-2, 603284
CCN2	100%	100%	100%	100%	99.7%	?Spondyloepimetaphyseal dysplasia, Li-Shao-Li type, 621099;Kyphomelic dysplasia, 211350
CCN6	100%	100%	100%	100%	99.7%	Progressive pseudorheumatoid dysplasia, 208230
CCNB3	100%	100%	99.5%	92.1%	73.8%	
CCND2	100%	100%	100%	100%	99.3%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNF	100%	100%	100%	100%	98.9%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141
CCNK	100%	100%	99.9%	98.9%	95.1%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CCNO	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 29, 615872
CCNP	100%	100%	100%	100%	99.4%	
CCNQ	96.7%	96.7%	99.5%	90.1%	70.1%	STAR syndrome, 300707

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CCR2	100%	100%	100%	100%	99.9%	{HIV infection, susceptibility/resistance to}, 609423;Polycystic lung disease, 219600
CCT2	97.1%	94.6%	100%	100%	99.9%	
CCT3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with speech or visual impairment and brain hypomyelination, 621034
CCT5	100%	100%	100%	100%	99.5%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CCT6A	100%	100%	100%	99.9%	99.6%	
CD151	100%	100%	100%	100%	99.7%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CD164	100%	100%	100%	100%	100%	?Deafness, autosomal dominant 66, 616969
CD19	100%	100%	100%	100%	99%	Immunodeficiency, common variable, 3, 613493
CD247	77.2%	71.4%	100%	100%	99.5%	?Immunodeficiency 25, 610163
CD27	100%	100%	100%	100%	99.7%	Lymphoproliferative syndrome 2, 615122
CD274	100%	100%	100%	100%	100%	?Autoimmune disease, multisystem, infantile-onset, 5, 621235
CD28	100%	100%	100%	100%	99.9%	?Immunodeficiency 123 with HPV-related verrucosis, 620901
CD2AP	100%	100%	100%	100%	99.8%	Glomerulosclerosis, focal segmental, 3, 607832
CD320	73.6%	73.6%	100%	100%	99.3%	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CD36	100%	100%	100%	100%	99.8%	Platelet glycoprotein IV deficiency, 608404;{Coronary heart disease, susceptibility to, 7}, 610938;{Malaria, cerebral, susceptibility to}, 611162;{Malaria, cerebral, reduced risk of}, 611162
CD3D	100%	100%	100%	100%	99.7%	Immunodeficiency 19, severe combined, 615617
CD3E	100%	100%	100%	100%	99.8%	Immunodeficiency 18, 615615;Immunodeficiency 18, SCID variant, 615615
CD3G	100%	100%	100%	100%	99.7%	Immunodeficiency 17, CD3 gamma deficient, 615607
CD4	100%	100%	100%	100%	99.6%	Immunodeficiency 79, 619238;OKT4 epitope deficiency, 613949
CD40	100%	100%	100%	100%	99.2%	Immunodeficiency with hyper-IgM, type 3, 606843
CD40LG	100%	100%	99.7%	93.5%	75%	Immunodeficiency, X-linked, with hyper-IgM, 308230
CD46	100%	100%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD48	100%	100%	100%	100%	99.7%	
CD55	96.6%	93.7%	100%	100%	99.5%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CD59	89.3%	89.3%	100%	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CD70	100%	100%	100%	100%	99.1%	Lymphoproliferative syndrome 3, 618261
CD79A	100%	100%	100%	100%	98.9%	Agammaglobulinemia 3, 613501
CD79B	100%	100%	100%	100%	99.5%	Agammaglobulinemia 6, 612692
CD81	100%	99.6%	100%	100%	99.4%	Immunodeficiency, common variable, 6, 613496
CD8A	100%	100%	100%	100%	99.5%	Immunodeficiency 116, 608957
CD96	100%	100%	100%	100%	99.8%	C syndrome, 211750
CDAN1	100%	100%	100%	100%	98.9%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDC14A	96.9%	96.9%	100%	100%	99.8%	Deafness, autosomal recessive 32, with or without immotile sperm, 608653
CDC20	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 14, 620276
CDC23	100%	100%	100%	100%	99.8%	
CDC40	100%	100%	100%	100%	99.6%	?Pontocerebellar hypoplasia, type 15, 619302
CDC42	100%	100%	100%	100%	99.6%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100%	100%	100%	100%	99.5%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841
CDC45	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 7, 617063
CDC6	100%	100%	100%	100%	99.4%	Meier-Gorlin syndrome 5, 613805

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CDC73	100%	100%	100%	99.9%	99.4%	Hyperparathyroidism, familial primary, 145000;Parathyroid adenoma with cystic changes, 145001;Parathyroid carcinoma, 608266;Hyperparathyroidism-jaw tumor syndrome, 145001
CDCA7	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910
CDH1	100%	100%	100%	100%	99.6%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480
CDH11	100%	100%	100%	100%	99.9%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380
CDH15	100%	100%	100%	99.9%	99%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	100%	100%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CDH23	100%	100%	100%	100%	99.4%	Usher syndrome, type 1D, 601067;{Pituitary adenoma 5, multiple types}, 617540;Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 12, 601386
CDH3	100%	100%	100%	100%	99.6%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDH4	100%	100%	100%	100%	99.6%	
CDHR1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 613660;Cone-rod dystrophy 15, 613660;Retinitis pigmentosa 65, 613660
CDIN1	100%	100%	100%	100%	99.7%	Dyserythropoietic anemia, congenital, type Ib, 615631
CDK10	100%	100%	100%	100%	99.6%	Al Kaissi syndrome, 617694
CDK13	100%	100%	100%	100%	99.1%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK16	100%	100%	98.5%	87.5%	69.3%	
CDK19	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 87, 618916
CDK4	100%	100%	100%	100%	99.6%	{Melanoma, cutaneous malignant, 3}, 609048;Microcephaly 31, primary, autosomal recessive, 621507

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CDK5	100%	100%	100%	100%	99.4%	?Lissencephaly 7 with cerebellar hypoplasia, 616342
CDK5RAP2	100%	100%	100%	100%	99.7%	Microcephaly 3, primary, autosomal recessive, 604804
CDK6	100%	100%	100%	100%	99%	?Microcephaly 12, primary, autosomal recessive, 616080
CDK8	92.4%	92.4%	100%	100%	99.6%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDK9	100%	100%	100%	100%	99.2%	
CDKL5	95.7%	95.7%	98.8%	89.3%	70.8%	Developmental and epileptic encephalopathy 2, 300672
CDKN1A	100%	100%	100%	100%	99.8%	
CDKN1B	100%	100%	100%	100%	99.6%	Multiple endocrine neoplasia, type IV, 610755
CDKN1C	100%	100%	100%	100%	99%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDKN2A	100%	100%	100%	99.8%	98.5%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100%	100%	100%	100%	99.5%	
CDKN2C	100%	100%	100%	100%	98.7%	
CDON	100%	100%	100%	100%	99.9%	Holoprosencephaly 11, 614226
CDSN	100%	100%	100%	100%	99.5%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CDT1	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 4, 613804
CDY1	50%	50%	47%	40.1%	26.1%	
CDY1B	50%	50%	47.1%	37.6%	24.5%	
CDY2A	50%	50%	49.4%	42.8%	26.8%	
CDY2B	50%	50%	48.9%	43.5%	27.5%	
CEACAM16	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 4B, 614614;Deafness, autosomal recessive 113, 618410
CEBPA	100%	100%	99.9%	98.8%	92.6%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626
CEBPE	100%	100%	100%	100%	99.4%	?Immunodeficiency 108 with autoinflammation, 260570;Specific granule deficiency, 245480
CEL	100%	100%	99.9%	99.4%	97.7%	Maturity-onset diabetes of the young, type VIII, 609812
CELA2A	100%	100%	100%	99.7%	98.6%	Abdominal obesity-metabolic syndrome 4, 618620
CELA3B	100%	100%	100%	99.8%	97.8%	
CELF2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 97, 619561
CELF4	100%	100%	100%	99.9%	99%	
CELSR1	100%	100%	100%	100%	99.2%	Lymphatic malformation 9, 619319;Yellow nail syndrome, 153300
CELSR3	100%	100%	100%	100%	99.4%	
CENATAC	100%	100%	100%	100%	99.4%	?Mosaic variegated aneuploidy syndrome 4, 620153

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CENPE	100%	100%	100%	99.9%	99.5%	?Microcephaly 13, primary, autosomal recessive, 616051
CENPF	98.6%	98.6%	100%	100%	99.8%	Stromme syndrome, 243605
CENPH	100%	100%	100%	100%	99.6%	
CENPT	95.4%	95.4%	100%	100%	99.6%	?Short stature and microcephaly with genital anomalies, 618702
CEP104	95.1%	95.1%	100%	100%	99.6%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988
CEP112	96.8%	96.8%	100%	100%	99.9%	Spermatogenic failure 44, 619044
CEP120	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP135	95.8%	95.8%	100%	100%	99.7%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100%	100%	100%	100%	99.9%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP162	100%	100%	100%	100%	99.8%	
CEP164	100%	100%	100%	100%	99.5%	Nephronophthisis 15, 614845
CEP19	100%	100%	100%	100%	99.7%	?Morbid obesity and spermatogenic failure, 615703
CEP250	100%	100%	100%	100%	99.5%	Cone-rod dystrophy and hearing loss 2, 618358

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CEP290	100%	100%	100%	100%	99.8%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP295	100%	100%	100%	100%	99.9%	Seckel syndrome 11, 620767
CEP41	100%	100%	100%	100%	99.8%	Joubert syndrome 15, 614464
CEP55	100%	100%	100%	100%	99.9%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100%	100%	100%	100%	99.7%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	92.8%	92.8%	100%	100%	99.7%	?Seckel syndrome 6, 614728
CEP78	100%	100%	100%	100%	99.6%	Cone-rod dystrophy and hearing loss, 617236
CEP83	100%	100%	100%	100%	99.8%	Nephronophthisis 18, 615862
CEP85L	100%	100%	100%	100%	99.7%	Lissencephaly 10, 618873
CEP89	98.3%	98.3%	100%	100%	99.6%	
CERKL	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 26, 608380
CERS1	100%	99.6%	100%	100%	99%	Epilepsy, progressive myoclonic, 8, 616230
CERS3	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 9, 615023

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CERT1	98.1%	98.1%	100%	99.9%	99.6%	Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies, 616351
CES1	99.9%	99.9%	99.8%	99.1%	97%	Drug metabolism, altered, CES1-related, 618057
CETP	83.8%	83.8%	100%	100%	99.8%	[High density lipoprotein cholesterol level QTL 10], 143470;Hyperalphalipoproteinemia, 143470
CFAP20	100%	100%	100%	100%	99.5%	
CFAP221	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 55, 279000
CFAP251	100%	100%	100%	100%	99.7%	Spermatogenic failure 33, 618152
CFAP276	100%	100%	100%	100%	99.7%	
CFAP298	89.6%	86.3%	100%	100%	99.5%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100%	100%	100%	100%	100%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100%	100%	100%	100%	99.3%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406
CFAP43	100%	100%	100%	100%	99.9%	Hydrocephalus, normal pressure, 1, 236690;Spermatogenic failure 19, 617592
CFAP44	100%	100%	100%	100%	99.8%	Spermatogenic failure 20, 617593
CFAP45	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CFAP46	100%	100%	100%	100%	99.4%	
CFAP47	100%	100%	99.4%	92.8%	75.9%	Spermatogenic failure, X-linked 3, 301059
CFAP52	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP54	100%	100%	100%	100%	99.9%	Spermatogenic failure 98, 621124;Ciliary dyskinesia, primary, 54, 621125
CFAP57	100%	100%	100%	100%	99.7%	Spermatogenic failure 95, 620917
CFAP58	100%	100%	100%	100%	99.7%	Spermatogenic failure 49, 619144
CFAP61	100%	100%	100%	100%	99.7%	Spermatogenic failure 84, 620409
CFAP65	100%	100%	100%	100%	99.2%	Spermatogenic failure 40, 618664
CFAP69	100%	100%	100%	100%	99.7%	Spermatogenic failure 24, 617959
CFAP70	100%	100%	100%	100%	99.7%	?Spermatogenic failure 41, 618670
CFAP74	100%	100%	100%	99.7%	98.6%	Ciliary dyskinesia, primary, 49, without situs inversus, 620197
CFAP91	100%	100%	100%	100%	99.7%	Spermatogenic failure 51, 619177
CFB	100%	100%	100%	100%	99.4%	Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CFD	100%	100%	100%	100%	98%	Complement factor D deficiency, 613912
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	98.3%	96.4%	91.7%	87.3%	76.3%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR2	76.4%	76.4%	100%	99.8%	99.5%	
CFHR3	98%	96.9%	93.2%	89.3%	78.5%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100%	100%	100%	99.6%	98.7%	
CFHR5	100%	100%	100%	100%	99.8%	Nephropathy due to CFHR5 deficiency, 614809
CFI	98.6%	95.9%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CFL2	100%	100%	100%	100%	100%	Nemaline myopathy 7, autosomal recessive, 610687
CFP	100%	100%	98.8%	88.3%	70.4%	Properdin deficiency, X-linked, 312060

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CFTR	100%	100%	100%	100%	99.6%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CGN	100%	100%	100%	100%	99.5%	
CHAMP1	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579
CHASERR						Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012
CHAT	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHCHD10	100%	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHCHD2	90.1%	90.1%	100%	100%	99.8%	Parkinson disease 22, autosomal dominant, 616710
CHCHD4	100%	100%	100%	99.9%	98.8%	
CHD1	100%	100%	100%	100%	99.9%	Pilarowski-Bjornsson syndrome, 617682

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CHD2	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100%	100%	100%	99.9%	99.1%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	100%	100%	100%	99.5%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100%	100%	100%	99.8%	98.7%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHD8	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHEK1	100%	100%	100%	99.9%	99.7%	Oocyte/zygote/embryo maturation arrest 21, 620610
CHEK2	100%	100%	100%	100%	99.6%	Prostate cancer, somatic, 176807;Osteosarcoma, somatic, 259500;Tumor predisposition syndrome 4, breast/prostate/colorectal, 609265
CHIT1	98.5%	94.9%	100%	100%	99.6%	[Chitotriosidase deficiency], 614122
CHKA	100%	100%	100%	100%	99.1%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023
CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHM	99.5%	98.5%	98.8%	92.3%	75.2%	Choroideremia, 303100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CHMP1A	100%	100%	100%	99.9%	99.2%	Pontocerebellar hypoplasia, type 8, 614961
CHMP2B	90.8%	88.1%	100%	100%	99.7%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CHMP4B	100%	100%	100%	100%	99.4%	Cataract 31, multiple types, 605387
CHN1	96.5%	96.5%	100%	100%	99.7%	Duane retraction syndrome 2, 604356
CHP1	100%	100%	100%	100%	99.5%	?Spastic ataxia 9, autosomal recessive, 618438
CHRDL1	100%	100%	99.3%	91.9%	74.5%	Megalocornea 1, X-linked, 309300
CHRM1	100%	100%	100%	100%	99.4%	
CHRM2	100%	100%	100%	100%	99.9%	
CHRM3	100%	100%	100%	100%	99.8%	Prune belly syndrome, 100100
CHRNA1	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290
CHRNA2	100%	100%	100%	100%	98.9%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA3	92%	92%	100%	99.9%	99.4%	{Lung cancer susceptibility 2}, 612052;Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT, 191800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CHRNA4	100%	100%	100%	99.7%	96.4%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB1	100%	100%	100%	99.8%	98.5%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRNB2	100%	100%	100%	100%	99.1%	Epilepsy, nocturnal frontal lobe, 3, 605375
CHRND	100%	100%	100%	100%	99.5%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323;Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 3B, fast-channel, 616322;?Myasthenic syndrome, congenital, 3A, slow-channel, 616321
CHRNE	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	100%	100%	100%	99.9%	99.6%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CHST11	100%	100%	100%	100%	99.7%	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits, 618167
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHST3	100%	100%	100%	100%	98.9%	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CHST6	100%	100%	100%	100%	99.6%	Macular corneal dystrophy, 217800
CHST8	100%	100%	100%	99.9%	99.1%	
CHSY1	100%	99.9%	100%	99.8%	99.1%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100%	100%	100%	100%	99.6%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630
CIAO1	100%	100%	100%	99.7%	98.8%	Multiple mitochondrial dysfunctions syndrome 10, 620960
CIB1	100%	100%	100%	100%	99.6%	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267
CIB2	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 48, 609439
CIBAR1	98.3%	92.7%	100%	100%	99.8%	?Polydactyly, postaxial, type A9, 618219
CIC	100%	100%	100%	99.9%	99%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIDEC	100%	100%	100%	100%	99.8%	?Lipodystrophy, familial partial, type 5, 615238

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CIITA	100%	100%	100%	100%	99.2%	{Rheumatoid arthritis, susceptibility to}, 180300;MHC class II deficiency 1, 209920
CILK1	100%	100%	100%	100%	99.8%	Cranioectodermal dysplasia 6, 621337;{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
CIP2A	100%	100%	100%	99.9%	99.5%	
CIROP	98.3%	93.9%	100%	100%	99.7%	Heterotaxy, visceral, 12, autosomal, 619702
CIROZ	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 14, autosomal, 621080
CISD2	100%	100%	100%	99.9%	99.4%	Wolfram syndrome 2, 604928
CIT	95.8%	95.8%	100%	100%	99.4%	Microcephaly 17, primary, autosomal recessive, 617090
CITED2	100%	100%	100%	99.8%	98.5%	Atrial septal defect 8, 614433;Ventricular septal defect 2, 614431
CIZ1	100%	100%	100%	100%	99.3%	
CKAP2L	100%	100%	100%	100%	99.7%	Filippi syndrome, 272440
CLASP1	100%	100%	100%	100%	99.6%	
CLCC1	98.6%	98.6%	100%	100%	99.7%	Retinitis pigmentosa 32, 609913
CLCF1	100%	100%	100%	100%	99.8%	Cold-induced sweating syndrome 2, 610313
CLCN1	100%	100%	100%	100%	99.6%	Myotonia congenita, recessive, 255700;Myotonia congenita, dominant, 160800;Myotonia levior, 160800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CLCN2	100%	99.1%	100%	100%	99.6%	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN3	100%	100%	100%	100%	99.8%	?Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100%	100%	98.5%	88.6%	71.1%	Raynaud-Claes syndrome, 300114
CLCN5	100%	100%	99.4%	92.1%	73.4%	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990;Hypophosphatemic rickets, 300554;Dent disease 1, 300009;Nephrolithiasis, type I, 310468
CLCN6	97.7%	97.7%	100%	100%	99.6%	Ceroid lipofuscinosis, neuronal, 15, 619173
CLCN7	97.5%	97.5%	100%	100%	99.7%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLCNKA	100%	100%	100%	100%	99.5%	Bartter syndrome, type 4b, digenic, 613090

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CLCNKB	98.6%	97.4%	100%	100%	99.5%	Bartter syndrome, type 3, 607364;Bartter syndrome, type 4b, digenic, 613090
CLDN1	100%	100%	100%	100%	99.9%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100%	100%	100%	100%	99.8%	HELIX syndrome, 617671
CLDN11	100%	100%	100%	99.9%	99%	Leukodystrophy, hypomyelinating, 22, 619328
CLDN14	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 29, 614035
CLDN16	95%	95%	100%	100%	99.8%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	100%	100%	100%	99.5%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLDN2	100%	100%	98.6%	92.3%	74.9%	?Azoospermia, obstructive, with nephrolithiasis, 301060
CLDN5	100%	100%	100%	100%	99.3%	
CLDN9	100%	100%	100%	100%	99.2%	Deafness, autosomal recessive 116, 619093
CLEC3B	100%	100%	100%	100%	99.9%	Macular dystrophy, retinal, 4, 619977
CLEC4D	100%	100%	100%	100%	99.9%	
CLEC7A	99.3%	95.3%	100%	100%	99.6%	Candidiasis, familial, 4, autosomal recessive, 613108;{Aspergillosis, susceptibility to}, 614079
CLIC2	87.2%	87.2%	99.7%	92.8%	74.4%	
CLIC5	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 103, 616042
CLIP1	100%	100%	100%	100%	99.8%	
CLMP	100%	100%	100%	100%	99.7%	Congenital short bowel syndrome, 615237

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CLN3	93.1%	93.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 3, 204200;Retinitis pigmentosa 101, 621548
CLN5	83%	83%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	98.2%	98.2%	100%	100%	99.7%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLPP	100%	100%	100%	100%	98.9%	Perrault syndrome 3, 614129
CLPX	100%	100%	100%	100%	99.8%	?Protoporphyria, erythropoietic, 2, 618015
CLRN1	100%	100%	100%	99.9%	99.1%	Usher syndrome, type 3A, 276902;Retinitis pigmentosa 61, 614180
CLRN2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 117, 619174
CLTC	97.3%	96%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 56, 617854

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CLTCL1	100%	100%	100%	100%	99.6%	
CLUAP1	100%	100%	100%	100%	99.5%	
CLXN	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 53, 620642
CMAS	100%	100%	100%	100%	99.9%	
CMIP	100%	100%	100%	100%	99.4%	
CMPK2	100%	100%	100%	100%	99.1%	Basal ganglia calcification, idiopathic, 10, autosomal recessive, 621018
CNBP	100%	100%	100%	100%	100%	Myotonic dystrophy 2, 602668
CNGA1	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 49, 613756
CNGA2	100%	99.9%	98%	87.1%	68.1%	
CNGA3	100%	100%	100%	100%	99.2%	Achromatopsia 2, 216900
CNGB1	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 45, 613767
CNGB3	100%	100%	100%	99.9%	99.5%	Achromatopsia 3, 262300
CNKSR2	100%	100%	99.2%	91.3%	72.7%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008
CNNM2	100%	100%	100%	100%	99.4%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
CNNM4	100%	100%	100%	100%	99.4%	Jalili syndrome, 217080
CNOT1	100%	100%	100%	100%	99.7%	Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CNOT2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100%	100%	100%	99.9%	98.8%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNOT9	98.4%	93.6%	100%	100%	99.9%	
CNP	100%	100%	100%	100%	99.6%	?Leukodystrophy, hypomyelinating, 20, 619071
CNPY3	99.1%	95.7%	100%	100%	99.3%	Developmental and epileptic encephalopathy 60, 617929
CNTN1	100%	100%	100%	100%	99.8%	Congenital myopathy 12, 612540
CNTN2	100%	99.9%	100%	99.9%	99.5%	Epilepsy, early-onset, 5, with or without developmental delay, 615400
CNTNAP1	100%	100%	100%	100%	99.1%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100%	100%	100%	100%	99.7%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA1	100%	100%	100%	100%	99.7%	
COA3	100%	100%	100%	100%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 14, 619058
COA5	69.5%	69.5%	100%	99.8%	98.1%	?Mitochondrial complex IV, deficiency, nuclear type 9, 616500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COA6	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 13, 616501
COA7	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387
COA8	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	100%	99.8%	99.1%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COCH	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 9, 601369;?Deafness, autosomal recessive 110, 618094
COG1	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIg, 611209
COG2	100%	100%	100%	100%	99.4%	?Congenital disorder of glycosylation, type IIq, 617395
COG3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIbb, 620546
COG4	97%	97%	100%	100%	99.3%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	96.8%	96.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100%	100%	100%	100%	99.9%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COG7	95.1%	95.1%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIh, 611182
COL10A1	100%	100%	100%	100%	99.4%	Metaphyseal chondrodysplasia, Schmid type, 156500
COL11A1	100%	100%	100%	100%	99.8%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL11A2	100%	100%	100%	100%	99.1%	Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL12A1	100%	100%	100%	100%	99.8%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470
COL13A1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 19, 616720
COL14A1	100%	100%	100%	100%	99.7%	
COL17A1	100%	100%	100%	100%	99.4%	Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL18A1	99.2%	98%	100%	99.9%	99.2%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880
COL1A1	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420
COL1A2	100%	100%	100%	100%	99.8%	Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL25A1	100%	100%	100%	100%	99.7%	Fibrosis of extraocular muscles, congenital, 5, 616219
COL27A1	100%	100%	100%	100%	99.4%	Steel syndrome, 615155

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL2A1	100%	100%	100%	100%	99.5%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;Spondylometaphyseal dysplasia, Algerian type, 184253;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;Osteoarthritis with mild chondrodysplasia, 604864;Stickler syndrome, type I, 108300;Platyspondylic skeletal dysplasia, Torrance type, 151210;Spondyloepiphyseal dysplasia, Stanescu type, 616583;Avascular necrosis of the femoral head, 608805;Legg-Calve-Perthes disease, 150600
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	98.6%	98.5%	100%	100%	99.3%	Brain small vessel disease 2A, autosomal dominant, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519;Brain small vessel disease 2B, autosomal recessive, 621414
COL4A3	100%	100%	100%	100%	99.6%	Alport syndrome 3A, autosomal dominant, 104200;Hematuria, benign familial, 2, 620320;Alport syndrome 3B, autosomal recessive, 620536
COL4A4	99.7%	98.9%	100%	100%	99.7%	Hematuria, familial benign, 1, 141200;Alport syndrome 2, autosomal recessive, 203780
COL4A5	100%	100%	99.2%	91.7%	73%	Alport syndrome 1, X-linked, 301050
COL4A6	100%	100%	98.9%	90%	71.4%	?Deafness, X-linked 6, 300914

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL5A1	100%	100%	100%	100%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329
COL5A2	100%	100%	100%	100%	99.8%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL6A1	100%	100%	100%	100%	99.4%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	100%	99.8%	100%	99.9%	99.2%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	100%	99.7%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COL6A5	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL7A1	100%	100%	100%	100%	99.4%	Nail disorder, nonsyndromic congenital, 8, 607523;Epidermolysis bullosa dystrophica, Bart type, 132000;Epidermolysis bullosa dystrophica inversa, 226600;Epidermolysis bullosa dystrophica, autosomal recessive, 226600;Epidermolysis bullosa, pretibial, 131850;Epidermolysis bullosa dystrophica, autosomal dominant, 131750;Transient bullous of the newborn, 131705;Epidermolysis bullosa pruriginosa, 604129;Epidermolysis bullosa dystrophica, localisata variant, 226600
COL8A2	100%	100%	100%	100%	98.7%	Corneal dystrophy, posterior polymorphous 2, 609140;Corneal dystrophy, Fuchs endothelial, 1, 136800
COL9A1	100%	100%	100%	100%	99.7%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	100%	100%	99.3%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284
COL9A3	100%	100%	100%	100%	99.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC10	95%	86.3%	100%	100%	99.8%	3MC syndrome 3, 248340

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COLEC11	100%	100%	100%	100%	99.6%	3MC syndrome 2, 265050
COLGALT1	96%	92.4%	100%	100%	99%	Brain small vessel disease 3, 618360
COLQ	100%	100%	100%	100%	99.6%	Myasthenic syndrome, congenital, 5, 603034
COMP	96.8%	96.3%	100%	99.9%	99.3%	Pseudoachondroplasia, 177170;Carpal tunnel syndrome 2, 619161;Epiphyseal dysplasia, multiple, 1, 132400
COMT	92.7%	88.6%	100%	100%	99.5%	[Catechol-O-methyltransferase activity, variation in], 621296
COPA	100%	100%	100%	100%	99.5%	{Autoinflammation and autoimmunity, systemic, with immune dysregulation 1}, 616414
COPB1	96.3%	96.3%	100%	99.9%	99.7%	Baralle-Macken syndrome, 619255
COPB2	96.7%	96.7%	100%	100%	99.8%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800
COPG1	96.6%	95.8%	100%	99.9%	99.3%	?Immunodeficiency 128, 620983
COPZ1	86.8%	86.2%	100%	100%	99.8%	Neutropenia, severe congenital, 12, autosomal recessive, 621439
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COQ4	96.3%	96.3%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ5	94.4%	94.4%	100%	100%	99.5%	?Coenzyme Q10 deficiency, primary, 9, 619028
COQ6	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 6, 614650
COQ7	100%	100%	100%	100%	99.5%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	89.1%	87.2%	100%	99.9%	99.5%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ8B	92.1%	92.1%	100%	100%	99.6%	Nephrotic syndrome, type 9, 615573
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
CORIN	97.6%	97.6%	100%	100%	99.8%	?Cardiomyopathy, familial hypertrophic, 30, atrial, 620734;Preeclampsia/eclampsia 5, 614595
CORO1A	100%	100%	100%	100%	99.7%	Immunodeficiency 8, 615401
COX10	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX11	100%	100%	100%	99.9%	98.7%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX14	100%	100%	100%	100%	100%	?Mitochondrial complex IV deficiency, nuclear type 10, 619053

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COX15	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	61.9%	61.9%	100%	99.8%	99.4%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX18	100%	100%	100%	99.9%	99.4%	?Mitochondrial complex IV deficiency, nuclear type 25, 621487;Charcot-Marie-Tooth disease, axonal, type 2MM, 621488
COX20	100%	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
COX4I1	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 16, 619060
COX4I2	100%	99.1%	100%	100%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX5A	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 20, 619064
COX5B	91.4%	80.9%	100%	100%	99.3%	
COX6A1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, recessive intermediate D, 616039
COX6A2	100%	100%	100%	99.9%	97.6%	Mitochondrial complex IV deficiency, nuclear type 18, 619062
COX6B1	80.9%	80.9%	100%	99.8%	98.7%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
COX6B2	100%	100%	100%	100%	99.4%	
COX6C	100%	100%	100%	100%	99.8%	
COX7A1	100%	100%	100%	100%	99.6%	
COX7A2	66.4%	66.4%	100%	100%	99.6%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COX7B	100%	100%	98.7%	86.9%	70%	Linear skin defects with multiple congenital anomalies 2, 300887
COX7B2	100%	100%	100%	99.9%	99.3%	
COX7C	78.1%	70.4%	100%	100%	99.6%	
COX8A	100%	100%	100%	99.7%	97.3%	?Mitochondrial complex IV deficiency, nuclear type 15, 619059
COX8C	100%	100%	100%	100%	99.7%	
CP	100%	100%	100%	100%	99.9%	Aceruloplasminemia, 604290
CPA1	100%	100%	100%	100%	99.6%	
CPA6	100%	100%	100%	100%	99.8%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417
CPAMD8	100%	100%	100%	99.9%	99.3%	Anterior segment dysgenesis 8, 617319
CPAP	100%	100%	100%	100%	99.7%	Microcephaly 6, primary, autosomal recessive, 608393;Seckel syndrome 4, 613676
CPE	97.5%	97.5%	100%	99.9%	99.1%	BDV syndrome, 619326
CPLANE1	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CPLX1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 63, 617976
CPN1	100%	100%	100%	99.9%	99%	Carboxypeptidase N deficiency, 212070
CPOX	93.4%	93.4%	100%	99.9%	99%	Coproporphyria, 121300;Harderoporphyria, 618892

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CPS1	100%	100%	100%	100%	99.7%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF1	100%	100%	100%	99.9%	99%	Myopia 27, 618827
CPSF3	95.9%	95.9%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CPT1A	100%	100%	100%	100%	99.5%	CPT deficiency, hepatic, type IA, 255120
CPT1C	100%	100%	100%	100%	99.3%	?Spastic paraplegia 73, autosomal dominant, 616282
CPT2	97.3%	96.3%	100%	100%	99.7%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110
CR2	100%	100%	100%	100%	99.9%	{Systemic lupus erythematosus, susceptibility to, 9}, 610927;?Immunodeficiency, common variable, 7, 614699
CRACR2A	100%	100%	100%	100%	99.6%	
CRADD	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRAT	99.2%	96.9%	100%	99.8%	99.1%	?Neurodegeneration with brain iron accumulation 8, 617917

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CRB1	98.6%	98.6%	100%	100%	99.9%	Leber congenital amaurosis 8, 613835;Retinitis pigmentosa-12, 600105;Pigmented paravenous chorioretinal atrophy, 172870
CRB2	100%	100%	100%	100%	99.6%	Focal segmental glomerulosclerosis 9, 616220;Ventriculomegaly with cystic kidney disease, 219730
CRBN	100%	99.6%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREB1	100%	100%	100%	100%	99.7%	Histiocytoma, angiomatoid fibrous, somatic, 612160
CREB3	100%	100%	100%	99.9%	99.4%	
CREB3L1	100%	100%	100%	100%	99.2%	Osteogenesis imperfecta, type XVI, 616229
CREB3L3	100%	100%	100%	100%	99.5%	Hypertriglyceridemia 2, 619324
CREBBP	100%	100%	100%	99.9%	99.5%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRELD1	100%	100%	100%	100%	99.6%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	100%	100%	100%	100%	99.9%	Rothmund-Thomson syndrome, type 3, 615789
CRIPTO	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CRLF1	94.8%	90.1%	99.7%	98.1%	93.3%	Cold-induced sweating syndrome 1, 272430
CRLS1	89.6%	84.4%	100%	99.7%	99.4%	Combined oxidative phosphorylation deficiency 57, 620167
CRNKL1	100%	100%	100%	100%	99.8%	Microcephaly, progressive, with simplified gyral pattern and cerebellar hypoplasia, 621436
CRPPA	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CRTAP	100%	100%	100%	100%	99.2%	Osteogenesis imperfecta, type VII, 610682
CRTC1	99%	96.7%	100%	100%	99.1%	Mucoepidermoid salivary gland carcinoma
CRX	100%	100%	100%	100%	99.1%	Leber congenital amaurosis 7, 613829;Cone-rod retinal dystrophy-2, 120970
CRYAA	100%	100%	100%	100%	99.3%	Cataract 9, multiple types, 604219
CRYAB	100%	100%	100%	100%	99.7%	Myopathy, myofibrillar, 2B, infantile-onset, 613869;Myopathy, myofibrillar, 2A, adult-onset, 608810;Cataract 16, multiple types, 613763;Cardiomyopathy, dilated, 1II, 615184
CRYBA1	100%	100%	100%	100%	99.7%	Cataract 10, multiple types, 600881
CRYBA2	100%	100%	100%	99.9%	98.7%	?Cataract 42, 115900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CRYBA4	100%	100%	100%	100%	99.8%	Cataract 23, 610425
CRYBB1	100%	100%	100%	100%	99.5%	Cataract 17, multiple types, 611544
CRYBB2	100%	100%	100%	100%	99.9%	Cataract 3, multiple types, 601547
CRYBB3	100%	100%	100%	100%	99.6%	Cataract 22, 609741
CRYGB	100%	100%	100%	100%	99.6%	Cataract 39, multiple types, autosomal dominant, 615188
CRYGC	100%	100%	100%	100%	99.3%	Cataract 2, multiple types, 604307
CRYGD	100%	100%	100%	99.9%	99.3%	Cataract 4, multiple types, 115700
CRYGS	100%	100%	100%	100%	99.8%	Cataract 20, multiple types, 116100
CRYL1	100%	100%	100%	100%	99%	
CRYM	96.3%	93.1%	100%	100%	99.3%	Deafness, autosomal dominant 40, 616357
CSDE1	97.8%	97.8%	100%	100%	99.8%	
CSF1R	97.3%	97.3%	100%	100%	99.6%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSF2RA	46.6%	45%	50%	50%	49.5%	Surfactant metabolism dysfunction, pulmonary, 4, 300770
CSF2RB	100%	100%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 5, 614370
CSF3R	100%	100%	100%	100%	99.5%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CSGALNACT1	100%	100%	100%	100%	99.9%	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CSMD1	100%	100%	100%	99.9%	99.4%	
CSNK1D	100%	100%	100%	100%	99.4%	Advanced sleep-phase syndrome, familial, 2, 615224
CSNK1G1	100%	100%	100%	100%	99.7%	
CSNK2A1	94.2%	94.2%	100%	100%	99.7%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100%	100%	100%	100%	99.7%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	96.9%	96.9%	100%	100%	99.6%	Joubert syndrome 21, 615636
CSRP3	100%	100%	100%	100%	99.9%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
CST3	100%	100%	100%	99.7%	96.9%	{Macular degeneration, age-related, 11}, 611953;Leukodystrophy, adult-onset, autosomal dominant, without amyloid angiopathy, 621214;Cerebral amyloid angiopathy, 105150
CST6	100%	100%	100%	99.8%	98.6%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100%	100%	100%	100%	100%	Peeling skin syndrome 4, 607936
CSTB	100%	100%	100%	100%	99.4%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CSTF2	98.6%	95.9%	98.9%	91.7%	74.4%	?Intellectual developmental disorder, X-linked 113, 301116
CT55	100%	100%	99.1%	92.5%	74%	?Spermatogenic failure, X-linked, 7, 301106

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CTBP1	100%	99.6%	100%	99.8%	99.3%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	90.8%	90.5%	100%	100%	99.2%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTF1	100%	100%	100%	99.6%	96.8%	
CTH	100%	100%	100%	100%	100%	Cystathioninuria, 219500
CTHRC1	100%	100%	100%	100%	99.5%	Barrett esophagus/esophageal adenocarcinoma, 614266
CTLA4	93.2%	93.2%	100%	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CTNNA1	94.8%	94.8%	100%	100%	99.8%	Macular dystrophy, patterned, 2, 608970
CTNNA2	100%	100%	100%	100%	99.6%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNA3	100%	100%	100%	100%	100%	Arrhythmogenic right ventricular dysplasia 13, 615616

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CTNNB1	100%	100%	100%	100%	99.9%	Exudative vitreoretinopathy 7, 617572;Pilomatricoma, somatic, 132600;Colorectal cancer, somatic, 114500;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075;Medulloblastoma, somatic, 155255;Ovarian cancer, somatic, 167000;Hepatocellular carcinoma, somatic, 114550
CTNNBL1	100%	100%	100%	100%	99.7%	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846
CTNND1	100%	100%	100%	100%	99.9%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100%	100%	100%	99.9%	98.4%	
CTNS	100%	100%	100%	100%	99.4%	Cystinosis, nephropathic, 219800;Cystinosis, ocular nonnephropathic, 219750;Cystinosis, late-onset juvenile or adolescent nephropathic, 219900;Cystinosis, atypical nephropathic, 219800
CTPS1	100%	100%	100%	100%	99.7%	Immunodeficiency 24, 615897
CTR9	100%	100%	100%	100%	99.6%	
CTRC	100%	100%	100%	100%	99.6%	{Pancreatitis, chronic, susceptibility to}, 167800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CTSA	100%	100%	100%	100%	99.5%	Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394
CTSB	87.2%	84.5%	100%	100%	99.4%	Keratolytic winter erythema, 148370
CTSC	94.8%	94.8%	100%	100%	99.8%	Periodontitis 1, aggressive, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	100%	99.9%	99.4%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CTSH	93.4%	92.1%	100%	100%	99.3%	
CTSK	100%	99.8%	100%	100%	99.7%	Pycnodysostosis, 265800
CTSZ	81.3%	73.5%	100%	99.9%	99.6%	
CTTNBP2	100%	100%	100%	100%	99.8%	
CTU2	100%	100%	100%	100%	99.2%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUBN	100%	100%	100%	100%	99.8%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CUL3	97.6%	97.6%	100%	100%	99.8%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CUL4B	96.7%	96.7%	99.2%	91.3%	74%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUL7	100%	100%	100%	100%	99.4%	3-M syndrome 1, 273750
CUX1	100%	100%	100%	99.8%	98.8%	Neurodevelopmental disorder with developmental delay and with or without motor or speech delay, 618330
CUX2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 67, 618141
CWC27	82.6%	82.6%	100%	100%	99.8%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXCR2	100%	100%	100%	99.9%	99.2%	?WHIM syndrome 2, 619407
CXCR4	99%	99%	100%	100%	99.5%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYB561	100%	100%	100%	100%	99.2%	Orthostatic hypotension 2, 618182
CYB5A	57.7%	57.7%	100%	100%	99.9%	Methemoglobinemia and ambiguous genitalia, 250790
CYB5R3	95.7%	95%	100%	100%	99.2%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYBA	69.7%	69.3%	100%	100%	99.3%	Chronic granulomatous disease 4, autosomal recessive, 233690

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CYBB	100%	100%	99.4%	91.3%	73.7%	Immunodeficiency 34, mycobacteriosis, X-linked, 300645;Chronic granulomatous disease, X-linked, 306400
CYBC1	100%	100%	100%	100%	99.6%	Chronic granulomatous disease 5, autosomal recessive, 618935
CYBRD1	100%	100%	100%	100%	99.6%	
CYC1	96.9%	91.9%	100%	100%	99.3%	Mitochondrial complex III deficiency, nuclear type 6, 615453
CYCS	100%	100%	100%	100%	99.9%	Thrombocytopenia 4, 612004
CYFIP2	96.2%	96.2%	100%	100%	99.6%	Developmental and epileptic encephalopathy 65, 618008
CYLC1	100%	100%	99.3%	91.5%	74.3%	{Spermatogenic failure, X-linked, 8, susceptibility to}, 301119
CYLD	100%	100%	100%	100%	99.9%	Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP11A1	100%	98%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	100%	99.4%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CYP11B2	100%	100%	100%	100%	99.3%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Aldosterone to renin ratio raised;{Low renin hypertension, susceptibility to};Hypoaldosteronism, congenital, due to CMO II deficiency, 610600
CYP17A1	100%	100%	100%	100%	99.5%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546
CYP1B1	100%	100%	100%	100%	99.5%	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300;Anterior segment dysgenesis 6, multiple subtypes, 617315
CYP21A2	100%	100%	100%	100%	99.4%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
CYP24A1	100%	100%	100%	100%	99.6%	Hypercalcemia, infantile, 1, 143880
CYP26B1	100%	100%	100%	100%	99.3%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
CYP26C1	100%	100%	100%	100%	99.4%	Focal facial dermal dysplasia 4, 614974
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP27B1	100%	100%	100%	100%	99.6%	Vitamin D-dependent rickets, type I, 264700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CYP2A6	100%	100%	99.9%	98.8%	95.9%	{Tagafur, poor metabolism of}, 621426;{Lung cancer, resistance to}, 211980;Coumarin resistance, 122700;{Nicotine addiction, protection from}, 188890
CYP2B6	100%	100%	100%	100%	99.8%	{Efavirenz central nervous system toxicity, susceptibility to}, 614546;Efavirenz, poor metabolism of, 614546
CYP2C19	87.8%	87.7%	100%	100%	99.9%	Proguanil poor metabolizer, 609535;Mephenytoin poor metabolizer, 609535;Clopidogrel, impaired responsiveness to, 609535;Omeprazole poor metabolizer, 609535
CYP2C8	92.3%	92.3%	100%	100%	99.9%	{Drug metabolism, altered, CYP2C8-related}, 618018
CYP2C9	100%	100%	100%	100%	99.6%	Warfarin sensitivity, 122700;Tolbutamide poor metabolizer
CYP2R1	100%	100%	100%	100%	99.5%	Rickets due to defect in vitamin D 25-hydroxylation deficiency, 600081
CYP2U1	100%	100%	100%	99.9%	98.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP3A4	100%	100%	100%	100%	99.9%	Vitamin D-dependent rickets, type 3, 619073
CYP4F22	100%	100%	100%	100%	99.1%	Ichthyosis, congenital, autosomal recessive 5, 604777

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CYP4V2	100%	100%	100%	100%	99.8%	Bietti crystalline corneoretinal dystrophy, 210370
CYP7A1	100%	100%	100%	100%	99.8%	
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812
D2HGDH	99.9%	98.4%	100%	99.9%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAAM2	100%	100%	100%	100%	99.5%	Nephrotic syndrome, type 24, 619263
DAB1	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 37, 615945
DACT1	100%	100%	100%	100%	99.5%	Townes-Brocks syndrome 2, 617466
DAG1	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAGLA	100%	100%	100%	100%	99.3%	Neuroocular syndrome 2, paroxysmal type, 168885
DALRD3	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 86, 618910
DAND5	78.8%	78.8%	100%	99.9%	99.1%	Heterotaxy, visceral, 13, autosomal, 621079
DAO	100%	100%	100%	100%	99.5%	
DAP3	91.7%	90.8%	100%	100%	99.6%	Perrault syndrome 7, 621101
DARS1	100%	100%	100%	100%	99.8%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485
DAW1	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 52, 620570
DAZ1	50%	50%	48%	39.4%	27%	
DAZ2	50%	50%	46.6%	39.6%	26.7%	
DAZ3	50%	49.9%	41.7%	31.8%	21.8%	
DAZ4	49.8%	49.5%	42.7%	32.8%	20.5%	
DBF4	98.5%	97.5%	100%	99.9%	99.3%	
DBH	100%	100%	100%	100%	99.5%	Orthostatic hypotension 1, due to DBH deficiency, 223360
DBR1	100%	100%	100%	100%	99.6%	Xerosis and growth failure with immune and pulmonary dysfunction syndrome, 620510;{Encephalitis, acute, infection (viral)-induced, susceptibility to, 11}, 619441
DBT	100%	100%	100%	100%	99.9%	Maple syrup urine disease, type II, 620699
DCAF12L1	100%	100%	98.7%	89.7%	68.9%	
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCAF8	96%	96%	100%	100%	99.7%	?Giant axonal neuropathy 2, autosomal dominant, 610100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DCC	100%	100%	100%	100%	99.7%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCDC1	100%	100%	100%	100%	99.8%	
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DCHS1	100%	100%	100%	100%	99.6%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DCLRE1B	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 8, 620133
DCLRE1C	97.1%	97.1%	100%	100%	99.9%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DCN	89%	89%	100%	100%	99.8%	Corneal dystrophy, congenital stromal, 610048
DCPS	100%	100%	100%	100%	99.6%	Al-Raqad syndrome, 616459
DCT	100%	100%	100%	99.8%	98.9%	Oculocutaneous albinism, type VIII, 619165
DCTN1	100%	100%	100%	100%	99%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DCTN2	100%	100%	100%	100%	99.5%	
DCX	100%	100%	99%	89.8%	73%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DCXR	100%	100%	100%	100%	99.1%	[Pentosuria], 260800
DDB1	92.4%	92.3%	100%	100%	99.6%	White-Kernohan syndrome, 619426
DDB2	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DDC	100%	100%	100%	100%	99.7%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	100%	100%	100%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDOST	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type Ir, 614507
DDR1	100%	100%	100%	99.9%	99.5%	
DDR2	100%	100%	100%	100%	99.8%	Warburg-Cinotti syndrome, 618175;Spondylometaepiphyseal dysplasia, short limb-hand type, 271665
DDRGK1	100%	100%	100%	100%	99.6%	Spondyloepimetaphyseal dysplasia, Shohat type, 602557
DDX11	100%	100%	100%	100%	99.5%	Warsaw breakage syndrome, 613398
DDX17	100%	100%	100%	100%	99.5%	
DDX23	100%	100%	100%	99.9%	99.7%	
DDX39B	100%	100%	100%	100%	99.6%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DDX3X	99.4%	98.7%	99.1%	90.6%	72.1%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DDX3Y	50%	50%	48.5%	39.7%	21.3%	
DDX41	100%	100%	100%	100%	99.2%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DDX59	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome V, 174300
DDX6	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	93.3%	88.8%	100%	99.9%	98%	Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEF6	100%	100%	100%	100%	99%	Immunodeficiency 87 and autoimmunity, 619573
DEGS1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 49, 617281
DENND5B	96%	96%	100%	100%	99.7%	
DEPDC5	100%	100%	100%	99.9%	99.4%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DES	92.6%	92.6%	100%	99.9%	98.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DGAT1	100%	100%	100%	100%	99.6%	Diarrhea 7, protein-losing enteropathy type, 615863
DGAT2	100%	100%	100%	100%	99.5%	
DGCR8	95.6%	95.6%	100%	100%	99.3%	
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DGUOK	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 1, 617068;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070;Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880
DHCR24	97.8%	97.8%	100%	99.7%	98.6%	Desmosterolosis, 602398
DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	74.4%	74.4%	100%	100%	99.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DHFR	100%	100%	100%	100%	99.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHH	100%	100%	100%	100%	99.7%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHODH	89%	86.1%	100%	100%	99.5%	Miller syndrome, 263750
DHPS	96.4%	93.1%	100%	99.9%	99%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHRS3	100%	100%	100%	100%	99.5%	Craniosynostosis-scoliosis syndrome, 621499
DHRSX	50%	50%	50%	50%	49.7%	Congenital disorder of glycosylation, type 1DD, 301133
DHTKD1	92.7%	92.7%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX16	100%	100%	100%	100%	99.4%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with variable motor and speech impairment, 617804
DHX37	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DHX38	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 84, 618220
DHX9	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 75, 620988
DIABLO	90.8%	90.8%	100%	100%	99.4%	Deafness, autosomal dominant 64, 614152
DIAPH1	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIAPH2	100%	100%	99.3%	92.5%	75.5%	?Premature ovarian failure 2A, 300511
DIAPH3	100%	100%	100%	100%	99.8%	Auditory neuropathy, autosomal dominant 1, 609129
DICER1	100%	100%	100%	100%	99.8%	Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295
DIO1	100%	100%	100%	100%	99.9%	Thyroid hormone metabolism, abnormal, 2, 619855
DIP2B	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIP2C	100%	100%	100%	100%	99.4%	
DIS3	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DIS3L2	100%	100%	100%	99.9%	99.5%	Perlman syndrome, 267000
DISP1	100%	100%	100%	100%	99.8%	Holoprosencephaly 10, 621143
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLAT	100%	100%	100%	100%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLC1	100%	100%	100%	100%	99.6%	Colorectal cancer, somatic, 114500
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	100%	100%	98.7%	89.2%	70.2%	Intellectual developmental disorder, X-linked 90, 300850
DLG4	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLG5	100%	100%	100%	100%	99.5%	Yuksel-Vogel-Bauser syndrome, 620703
DLGAP5	99.9%	99.2%	100%	99.9%	99.5%	
DLK1	100%	100%	100%	99.9%	98.9%	
DLL1	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DLL3	100%	100%	100%	100%	98.8%	Spondylocostal dysostosis 1, autosomal recessive, 277300
DLL4	100%	100%	100%	100%	99.2%	Adams-Oliver syndrome 6, 616589

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DLST	100%	100%	100%	99.9%	99.5%	Pheochromocytoma/paraganglioma syndrome 7, 618475
DLX3	100%	100%	100%	99.9%	99.4%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	100%	100%	99.3%	?Orofacial cleft 15, 616788
DLX5	100%	100%	100%	100%	99.2%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DLX6	100%	100%	100%	99.2%	95.5%	
DMAC1	100%	100%	100%	100%	99.6%	
DMAC2	100%	100%	100%	100%	99.4%	
DMAC2L	100%	100%	100%	100%	99.7%	
DMAP1	92.4%	92.4%	100%	100%	99.7%	
DMC1	94.7%	94.7%	100%	100%	99.9%	
DMD	99.6%	99.6%	99.2%	91.5%	73.3%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DMGDH	98.8%	98.5%	100%	100%	99.8%	Dimethylglycine dehydrogenase deficiency, 605850
DMP1	100%	100%	100%	100%	99.1%	Hypophosphatemic rickets, AR, 241520
DMPK	100%	100%	100%	99.9%	99.1%	Myotonic dystrophy 1, 160900
DMRT1	100%	100%	100%	100%	99.5%	
DMRT2	100%	100%	100%	100%	99.7%	Spondylocostal dysostosis 7, autosomal recessive, 621523

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DMXL2	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DNA2	97.1%	97.1%	100%	100%	99.8%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156;Rothmund-Thomson syndrome, type 4, 620819;Seckel syndrome 8, 615807
DNAAF1	96.2%	95.9%	100%	100%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 19, 614935
DNAAF19	100%	100%	100%	100%	99.1%	Ciliary dyskinesia, primary, 17, 614679
DNAAF2	100%	100%	100%	99.9%	99.2%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100%	100%	100%	100%	99.9%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	99.9%	100%	99.9%	99.1%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	100%	99.5%	92.1%	76.8%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	100%	100%	99.4%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH10	100%	100%	100%	100%	99.7%	Spermatogenic failure 56, 619515

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DNAH11	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH12	100%	100%	100%	100%	99.7%	Spermatogenic failure 100, 621209
DNAH17	100%	100%	100%	99.9%	99.3%	Spermatogenic failure 39, 618643
DNAH2	100%	99.8%	100%	100%	99.3%	Spermatogenic failure 45, 619094
DNAH3	100%	100%	100%	100%	99.7%	
DNAH5	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH6	100%	100%	100%	100%	99.7%	
DNAH7	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 50, 620356
DNAH8	100%	100%	100%	100%	99.8%	Spermatogenic failure 46, 619095
DNAH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	95.7%	95.7%	100%	100%	99.8%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJA3	100%	100%	100%	100%	99.5%	
DNAJB11	100%	100%	100%	100%	99.8%	Polycystic kidney disease 6 with or without polycystic liver disease, 618061
DNAJB13	100%	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 34, 617091
DNAJB2	100%	100%	100%	99.9%	99.5%	Neuronopathy, distal hereditary motor, autosomal recessive 5, 614881

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DNAJB4	100%	100%	100%	100%	99.9%	Congenital myopathy 21 with early respiratory failure, 620326
DNAJB5	100%	100%	100%	100%	99.3%	
DNAJB6	92.2%	92.2%	100%	100%	99.9%	Muscular dystrophy, limb-girdle, autosomal dominant 1, 603511
DNAJC12	100%	100%	100%	100%	99.7%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DNAJC21	94.7%	94.7%	100%	100%	99.3%	Bone marrow failure syndrome 3, 617052
DNAJC3	92.1%	92.1%	100%	100%	99.8%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAJC30	100%	100%	100%	100%	99.7%	Leber-like hereditary optic neuropathy, autosomal recessive 1, 619382
DNAJC5	100%	100%	100%	100%	99.1%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350
DNAJC6	100%	100%	100%	99.9%	99.6%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528
DNAL1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 16, 614017
DNAL4	100%	100%	100%	99.8%	98%	?Mirror movements 3, 616059
DNALI1	100%	100%	100%	100%	99.4%	Spermatogenic failure 83, 620354

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DNASE1	100%	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to}, 152700
DNASE1L3	100%	100%	100%	100%	99.9%	Systemic lupus erythematosus 16, 614420
DNASE2	92.7%	92.7%	100%	100%	99.3%	Autoinflammatory-panc ytopenia syndrome, 619858
DNHD1	100%	100%	100%	100%	99.4%	Spermatogenic failure 65, 619712
DNM1	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopath y, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNM2	93.3%	91.7%	100%	99.9%	99.4%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368
DNMBP	100%	100%	100%	100%	99.6%	Cataract 48, 618415
DNMT1	100%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DNMT3A	100%	100%	100%	100%	99.5%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK11	100%	100%	99.3%	92.2%	74.3%	Autoinflammatory disease, multisystem, with immune dysregulation, X-linked, 301109
DOCK2	100%	100%	100%	100%	99.7%	Immunodeficiency 40, 616433
DOCK3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK4	100%	100%	100%	100%	99.7%	
DOCK6	100%	100%	100%	99.9%	99.3%	Adams-Oliver syndrome 2, 614219
DOCK7	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 23, 615859
DOCK8	98.6%	98.6%	100%	100%	99.7%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DOHH	98.4%	92.9%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DOK7	100%	100%	100%	99.9%	99.3%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
DOLK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Im, 610768
DONSON	100%	99.7%	100%	100%	99.7%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230;Meier-Gorlin syndrome 10, 621528
DOT1L	100%	100%	100%	100%	99.3%	Nil-Deshwar neurodevelopmental syndrome, 621265
DPAGT1	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPCD	93%	93%	100%	100%	99.3%	
DPF2	95.7%	95.7%	100%	99.8%	99.1%	Coffin-Siris syndrome 7, 618027
DPH1	100%	100%	100%	100%	99.3%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPH2	100%	100%	100%	100%	99.7%	Developmental delay with short stature, dysmorphic facial features, and sparse hair 2, 620062
DPH5	75.4%	75.4%	100%	100%	99.9%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DPM1	99.6%	96.9%	99.6%	98.4%	96.6%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	99.9%	100%	100%	99.5%	Congenital disorder of glycosylation, type Iu, 615042
DPM3	100%	100%	100%	100%	98.8%	?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15, 618992;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15, 612937
DPP6	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPP9	100%	100%	100%	99.9%	99.5%	Hatipoglu immunodeficiency syndrome, 620331
DPY19L2	100%	100%	100%	99.8%	99%	Spermatogenic failure 9, 613958
DPYD	86.9%	86.9%	100%	100%	99.9%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100%	100%	100%	100%	99.5%	Dihydropyrimidinuria, 222748
DPYSL2	100%	100%	100%	100%	99.4%	
DPYSL5	100%	100%	100%	99.9%	99.6%	Ritscher-Schinzel syndrome 4, 619435
DRAM2	98.4%	93.3%	100%	100%	99.7%	Cone-rod dystrophy 21, 616502
DRC1	100%	100%	100%	100%	99.5%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DRC3	96.8%	96.8%	100%	99.9%	99%	
DRD4	100%	100%	99.9%	99.6%	97.5%	{Attention deficit-hyperactivity disorder}, 143465;Autonomic nervous system dysfunction
DRG1	92.1%	92.1%	100%	100%	99.7%	Tan-Almurshedi syndrome, 620641
DRP2	100%	100%	98.8%	88.9%	69.3%	
DSC2	100%	100%	100%	99.9%	99.5%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	100%	100%	100%	100%	99.7%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	100%	100%	100%	100%	99.8%	Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG2	88%	88%	100%	100%	99.7%	Cardiomyopathy, dilated, 1BB, 612877;Arrhythmogenic right ventricular dysplasia 10, 610193
DSG3	100%	100%	100%	100%	99.8%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	100%	100%	100%	100%	99.8%	Hypotrichosis 6, 607903

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DSP	99.4%	99.2%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DSPP	100%	100%	99.1%	98.3%	97%	Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594
DST	100%	100%	100%	100%	99.7%	Congenital myopathy 29 with contractures, 621510;Neuropathy, hereditary sensory and autonomic, type VI, 614653;Lethal congenital contracture syndrome 12, 621511;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DSTYK	100%	100%	100%	100%	99.5%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DTNA	98.7%	97.2%	100%	100%	99.8%	Left ventricular noncompaction 1, with or without congenital heart defects, 604169;Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis 2, 620971
DTNBP1	96.4%	96.4%	100%	100%	99.7%	Hermansky-Pudlak syndrome 7, 614076
DTYMK	91.4%	84.2%	100%	100%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DUOX2	100%	100%	100%	99.9%	99.3%	Thyroid dyshormonogenesis 6, 607200
DUOXA2	100%	100%	100%	100%	99.1%	Thyroid dyshormonogenesis 5, 274900
DUSP1	100%	100%	100%	100%	99.6%	
DUSP6	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DUT	100%	100%	100%	99.8%	99%	Bone marrow failure and diabetes mellitus syndrome, 620044
DVL1	100%	100%	100%	99.9%	99.3%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	99.9%	99.1%	Robinow syndrome, autosomal dominant 3, 616894
DYM	100%	100%	100%	100%	99.9%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DYNC1H1	99.3%	99.3%	100%	99.9%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563
DYNC1I2	92.4%	92.4%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	95.7%	95.7%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	81.5%	81.4%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	58.6%	58.6%	100%	100%	99.3%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
DYRK1A	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
DYRK1B	96.2%	96.2%	100%	100%	99.6%	Abdominal obesity-metabolic syndrome 3, 615812

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

DYSF	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601;Miyoshi muscular dystrophy 1, 254130;Myopathy, distal, with anterior tibial onset, 606768
DZIP1	100%	100%	100%	100%	99.7%	Spermatogenic failure 47, 619102;?Mitral valve prolapse 3, 610840
DZIP1L	100%	100%	100%	100%	99.7%	Polycystic kidney disease 5, 617610
E2F1	100%	100%	100%	99.9%	99.1%	
EARS2	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100%	100%	100%	99.9%	99.2%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	100%	100%	98.8%	88.4%	69.5%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECE1	100%	100%	100%	100%	99.2%	{Hypertension, essential, susceptibility to}, 145500;?Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870
ECEL1	100%	100%	100%	100%	99.3%	Arthrogryposis, distal, type 5D, 615065
ECHS1	71.2%	69%	100%	100%	99.3%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
ECM1	100%	100%	100%	100%	99.3%	Urbach-Wiethe disease, 247100
ECSIT	100%	100%	100%	99.9%	99%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EDA	100%	100%	98.6%	87.3%	67.6%	Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	100%	100%	100%	99.7%	[Hair morphology 1, hair thickness], 612630;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	100%	100%	100%	100%	99.7%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDC3	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIv, 619493
EDN1	100%	100%	100%	100%	99.8%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706
EDN3	100%	100%	100%	99.9%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EDNRA	100%	100%	100%	100%	99.8%	{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367
EDNRB	100%	100%	100%	100%	99.5%	{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EED	99.1%	99.1%	100%	99.6%	97.9%	Cohen-Gibson syndrome, 617561
EEF1A2	99.4%	95.6%	100%	100%	99.1%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EEF1D	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with thin corpus callosum, hypotonia, and absent language, 621150
EEF2	100%	100%	100%	100%	99.4%	?Spinocerebellar ataxia 26, 609306
EEFSEC	100%	100%	100%	100%	98.7%	Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102
EFEMP1	84.9%	84.9%	100%	100%	99.8%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276
EFEMP2	99.6%	95.2%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IB, 614437

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EFHC1	97.7%	97.5%	100%	100%	99.7%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770
EFL1	96.3%	96.3%	100%	100%	99.9%	Shwachman-Diamond syndrome 2, 617941
EFNA4	100%	100%	100%	100%	99.4%	
EFNB1	100%	100%	98.8%	87.7%	69.3%	Craniofrontonasal dysplasia, 304110
EFNB2	100%	100%	100%	100%	99.6%	
EFTUD2	100%	100%	100%	100%	99.7%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EGF	100%	100%	100%	100%	99.8%	?Hypomagnesemia 4, renal, 611718
EGFLAM	100%	100%	100%	100%	99.8%	
EGFR	100%	100%	100%	100%	99.7%	Neonatal nephrocutaneous inflammatory syndrome, 616069;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980;Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980;{Nonsmall cell lung cancer, susceptibility to}, 211980
EGLN1	88%	88%	100%	99.9%	98.5%	Erythrocytosis, familial, 3, 609820;[Hemoglobin, high altitude adaptation], 609070
EGLN2	100%	100%	100%	100%	99.5%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EGR2	100%	100%	100%	100%	99.6%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253
EHD1	100%	100%	100%	100%	99.2%	
EHHADH	100%	100%	100%	100%	99.8%	?Fanconi renotubular syndrome 3, 615605
EHMT1	97.7%	97.7%	100%	100%	99.7%	Kleefstra syndrome 1, 610253
EHMT2	97%	97%	100%	99.9%	99%	
EIF1AX	100%	100%	98.9%	92.3%	73.7%	
EIF1AY	50%	50%	49.5%	40.3%	24%	
EIF2AK1	95%	94.1%	100%	100%	99.7%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100%	100%	100%	100%	99.8%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2AK3	100%	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
EIF2AK4	99%	99%	100%	100%	99.8%	Pulmonary venoocclusive disease 2, 234810
EIF2B1	93.5%	90.6%	100%	100%	99.8%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100%	100%	100%	99.9%	99.2%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EIF2B3	100%	100%	100%	100%	99.5%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	100%	99.9%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	100%	99.6%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF2S3	100%	100%	99.2%	91%	73.9%	MEHMO syndrome, 300148
EIF3A	100%	100%	100%	99.9%	99.5%	
EIF3B	100%	100%	100%	100%	99.2%	
EIF3F	100%	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A2	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455
EIF4A3	100%	100%	100%	100%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF4ENIF1	100%	100%	100%	100%	99.7%	
EIF5A	100%	100%	100%	100%	99.2%	Faundes-Banka syndrome, 619376
EIPR1	91.2%	91.2%	100%	100%	99.7%	
ELAC2	94.8%	94.8%	100%	99.9%	99.5%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ELANE	100%	100%	100%	99.9%	99%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
ELF2	100%	100%	100%	100%	99.7%	
ELF4	100%	100%	98.8%	88.6%	69.8%	Autoinflammatory syndrome, familial, X-linked, Behcet-like 2, 301074
ELFN1	100%	100%	100%	99.9%	98.5%	Dursun-Ozgul neurodevelopmental syndrome, 621344
ELMO2	97.9%	97.9%	100%	100%	99.6%	Vascular malformation, primary intraosseous, 606893
ELMOD3	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 88, 615429;?Deafness, autosomal dominant 81, 619500
ELN	100%	99.7%	100%	99.9%	99.1%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELOVL5	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia 38, 615957
ELP1	100%	100%	100%	100%	99.8%	{Medulloblastoma}, 155255;Dysautonomia, familial, 223900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ELP2	95%	94.7%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 58, 617270
ELP4	87.8%	87.8%	100%	100%	99.7%	?Aniridia 2, 617141
EMC1	99.6%	98.2%	100%	100%	99.6%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EMD	93.1%	91.5%	97.5%	87.2%	69.2%	Cardiomyopathy, dilated, 3C, 301163;Emery-Dreifuss muscular dystrophy 1, X-linked, 310300
EMG1	86.7%	86.7%	100%	99.9%	98.9%	Bowen-Conradi syndrome, 211180
EMILIN1	100%	99.7%	100%	100%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908
EML1	90.9%	90.9%	100%	100%	99.4%	Band heterotopia, 600348
EMP2	100%	100%	100%	100%	98.7%	Nephrotic syndrome, type 10, 615861
EMX2	100%	100%	100%	99.7%	96%	Schizencephaly, 269160
EN1	100%	100%	99.8%	99.3%	97.2%	?ENDOVE syndrome, limb-brain type, 619218
ENAM	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500
ENG	97.3%	96.8%	100%	99.9%	98.6%	Telangiectasia, hereditary hemorrhagic, type 1, 187300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ENO3	100%	100%	100%	100%	98.9%	Glycogen storage disease XIII, 612932
ENPP1	100%	100%	100%	100%	99.8%	{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
ENTPD1	100%	100%	100%	100%	99.6%	Spastic paraplegia 64, autosomal recessive, 615683
EOGT	95.8%	92.4%	100%	100%	99.6%	Adams-Oliver syndrome 4, 615297
EP300	100%	100%	100%	100%	99.5%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
EP400	100%	100%	100%	100%	99.4%	
EPAS1	100%	100%	100%	100%	99.6%	Erythrocytosis, familial, 4, 611783
EPB41	100%	100%	100%	100%	99.8%	Elliptocytosis-1, 611804
EPB41L1	99.9%	99.3%	100%	100%	99.4%	?Intellectual developmental disorder, autosomal dominant 11, 614257
EPB41L3	99.9%	99.3%	100%	100%	99.7%	
EPB42	100%	100%	100%	100%	99.5%	Spherocytosis, type 5, 612690
EPCAM	100%	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHA10	100%	100%	100%	99.9%	99%	?Deafness, autosomal dominant 88, 620283
EPHA2	100%	100%	100%	100%	99.5%	Cataract 6, multiple types, 116600
EPHA7	100%	100%	100%	100%	99.7%	
EPHB2	100%	100%	100%	100%	99.1%	?Bleeding disorder, platelet-type, 22, 618462;{Prostate cancer/brain cancer susceptibility, somatic}, 603688
EPHB4	97.7%	95.6%	100%	100%	99.3%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
EPHX1	91.3%	90.8%	100%	100%	99.5%	
EPHX2	100%	100%	100%	100%	99.7%	{Hypercholesterolemia, familial, due to LDLR defect, modifier of}, 143890
EPHX3	100%	100%	100%	100%	99.5%	
EPM2A	100%	100%	100%	99.6%	98.1%	Myoclonic epilepsy of Lafora 1, 254780
EPO	100%	100%	100%	100%	98.9%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
EPRS1	96.2%	96.2%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 15, 617951
EPS8	95.3%	95.3%	100%	100%	99.9%	?Deafness, autosomal recessive 102, 615974

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EPS8L2	100%	100%	100%	99.9%	99.1%	Deafness autosomal recessive 106, 617637
EPS8L3	100%	99.9%	100%	100%	99.3%	?Hypotrichosis 5, 612841
ERAL1	100%	100%	100%	99.9%	99.3%	Perrault syndrome 6, 617565
ERBB2	100%	100%	100%	100%	99.4%	Gastric cancer, somatic, 613659;Adenocarcinoma of lung, somatic, 211980;Ovarian cancer, somatic, 167000;?Visceral neuropathy, familial, 2, autosomal recessive, 619465;Glioblastoma, somatic, 137800
ERBB3	100%	100%	100%	99.9%	98.9%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERBB4	100%	100%	100%	100%	99.9%	Amyotrophic lateral sclerosis 19, 615515
ERBIN	100%	100%	100%	100%	99.8%	
ERCC1	100%	100%	100%	99.9%	99.2%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	96.3%	94.6%	100%	100%	99.3%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	100%	99.7%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ERCC4	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC5	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	100%	100%	99.6%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980
ERCC6L2	100%	100%	100%	100%	99.9%	Bone marrow failure syndrome 2, 615715
ERCC8	95.2%	95.2%	100%	99.9%	98.8%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERF	100%	100%	100%	100%	97.9%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERG	100%	100%	100%	100%	99.4%	Lymphatic malformation 14, 620602

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ERGIC1	88.5%	88.5%	100%	100%	99.1%	?Arthrogryposis multiplex congenita 2, neurogenic type, 208100
ERI1	100%	100%	100%	100%	99.6%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ERLIN1	100%	100%	100%	100%	99.8%	Spastic paraplegia 62, autosomal recessive, 615681
ERLIN2	100%	100%	100%	100%	99.5%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ERMARD	100%	100%	100%	100%	99.7%	?Periventricular nodular heterotopia 6, 615544
ESAM	99.7%	96.8%	100%	99.9%	99.1%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371
ESCO2	100%	100%	100%	100%	99.8%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
ESPN	100%	100%	100%	99.8%	98.2%	Deafness, autosomal recessive 36, 609006;Deafness, autosomal dominant 91, 621556;?Usher syndrome, type 1M, 618632
ESR1	100%	100%	100%	100%	99.7%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ESR2	100%	100%	100%	100%	99.8%	?Ovarian dysgenesis 8, 618187
ESRP1	100%	100%	100%	100%	99.7%	?Deafness, autosomal recessive 109, 618013
ESRRB	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 35, 608565
ETFA	82.4%	82.4%	100%	100%	99.6%	Glutaric acidemia IIA, 231680
ETFB	92.9%	92.9%	100%	100%	99.3%	Glutaric acidemia IIB, 231680
ETFDH	94.1%	92.3%	100%	100%	99.9%	Glutaric acidemia IIC, 231680
ETHE1	77.1%	77.1%	100%	100%	99.7%	Ethylmalonic encephalopathy, 602473
ETS1	100%	100%	100%	100%	99.7%	
ETV6	100%	100%	100%	100%	99.3%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EVC	100%	100%	100%	100%	99.5%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	100%	100%	99.7%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EWSR1	100%	100%	100%	99.6%	98%	Neuroepithelioma, 612219;Ewing sarcoma, 612219
EXOC2	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC3L2	100%	100%	100%	100%	98.9%	Brain malformation renal syndrome, 620943

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EXOC6B	97.1%	97.1%	100%	100%	99.7%	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395
EXOC7	98.6%	98.6%	100%	100%	99.6%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOC8	100%	100%	100%	100%	99.5%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXOSC1	100%	100%	100%	99.9%	99.4%	?Pontocerebellar hypoplasia, type 1F, 619304
EXOSC2	91.1%	91%	100%	100%	99.7%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	100%	98.6%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 1D, 618065
EXPH5	100%	100%	100%	100%	99.8%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
EXT1	100%	100%	100%	100%	99.5%	Exostoses, multiple, type 1, 133700;Chondrosarcoma, 215300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EXT2	100%	100%	100%	100%	99.6%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	100%	99.9%	100%	100%	99.7%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EYA1	100%	100%	100%	100%	100%	Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780
EYA4	100%	100%	100%	100%	99.9%	?Cardiomyopathy, dilated, 1J, 605362;Deafness, autosomal dominant 10, 601316
EYS	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 25, 602772
EZH1	100%	100%	100%	100%	99.7%	
EZH2	100%	100%	100%	100%	99.7%	Weaver syndrome, 277590
F10	100%	100%	100%	100%	99.4%	Factor X deficiency, 227600
F11	92.5%	92.5%	100%	100%	99.6%	Factor XI deficiency, autosomal dominant, 612416;Factor XI deficiency, autosomal recessive, 612416
F12	100%	100%	100%	100%	99.7%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

F13A1	100%	100%	100%	100%	99.7%	Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050
F13B	100%	100%	100%	100%	99.9%	Factor XIIIB deficiency, 613235
F2	90.8%	90.8%	100%	99.9%	99.4%	Hypoprothrombinemia, 613679;{Pregnancy loss, recurrent, susceptibility to, 2}, 614390;Dysprothrombinemia, 613679;Thrombophilia 1 due to thrombin defect, 188050;{Stroke, ischemic, susceptibility to}, 601367
F2RL3	100%	100%	100%	100%	99.5%	
F5	100%	100%	100%	100%	99.8%	Thrombophilia 2 due to activated protein C resistance, 188055;{Pregnancy loss, recurrent, susceptibility to, 1}, 614389;{Thrombophilia, susceptibility to, due to factor V Leiden}, 188055;{Budd-Chiari syndrome}, 600880;{Stroke, ischemic, susceptibility to}, 601367;Factor V deficiency, 227400
F7	99.5%	96.6%	100%	100%	99.6%	{Myocardial infarction, decreased susceptibility to}, 608446;Factor VII deficiency, 227500
F8	100%	100%	99.3%	92.3%	74.9%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071;Hemophilia A, 306700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

F9	100%	100%	99.7%	92.5%	75.6%	{Deep venous thrombosis, protection against}, 300807;Hemophilia B, 306900;Thrombophilia 8, X-linked, due to factor IX defect, 300807;{Warfarin sensitivity}, 301052
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAAH	100%	100%	100%	100%	99.6%	{Drug addiction, susceptibility to}, 606581
FAAP100	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group X, 621258
FAAP24	100%	100%	100%	100%	99.1%	
FADD	98.2%	92.7%	100%	99.9%	99.2%	Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction, 613759
FAH	95.2%	93.3%	100%	100%	99.5%	Tyrosinemia, type I, 276700
FAM111A	100%	100%	100%	100%	99.9%	Kenny-Caffey syndrome, type 2, 127000;Gracile bone dysplasia, 602361
FAM111B	100%	100%	100%	100%	99.4%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM149B1	100%	100%	100%	100%	99.9%	Joubert syndrome 36, 618763
FAM161A	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 28, 606068

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FAM177A1	86.5%	86.5%	100%	99.9%	99.3%	Neurodevelopmental disorder with white matter abnormalities and gait disturbance, 621152
FAM20A	100%	99.9%	100%	99.8%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20B	100%	100%	100%	100%	99.9%	
FAM20C	99.4%	97.2%	100%	100%	99.2%	Raine syndrome, 259775
FAM210A	100%	100%	100%	100%	99.6%	
FAM50A	93%	93%	98.9%	88.4%	67.1%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261
FAM83G	100%	100%	100%	100%	99.6%	
FAM83H	100%	100%	100%	100%	99.3%	
FAN1	95.9%	94%	100%	100%	99.6%	Interstitial nephritis, karyomegalic, 614817
FANCA	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group A, 227650
FANCB	96.2%	96.2%	99.4%	92.2%	74.7%	Fanconi anemia, complementation group B, 300514
FANCC	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	100%	99.7%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	100%	99.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	99.2%	Fanconi anemia, complementation group F, 603467

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FANCG	100%	99.5%	100%	100%	99.3%	Fanconi anemia, complementation group G, 614082
FANCI	100%	99.9%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053
FANCL	91.5%	89.3%	100%	100%	99.1%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAR1	91%	90.9%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSA	100%	100%	100%	100%	99.8%	?Rajab interstitial lung disease with brain calcifications 2, 619013
FARSB	94%	94%	100%	100%	99.7%	Rajab interstitial lung disease with brain calcifications 1, 613658
FAS	100%	100%	100%	100%	99.6%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FASLG	100%	100%	100%	99.9%	99.6%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
FASTKD2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 44, 618855
FASTKD5	100%	100%	100%	100%	99.9%	Mitochondrial complex IV deficiency, nuclear type 24, 621431
FAT1	100%	100%	100%	100%	99.7%	
FAT2	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 45, 617769
FAT3	100%	100%	100%	100%	99.8%	
FAT4	100%	100%	100%	99.9%	99.5%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN1	96.8%	94.6%	100%	99.9%	99.2%	
FBLN5	92.8%	92.8%	100%	99.9%	99.4%	Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FBN1	100%	100%	100%	100%	99.8%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	99.2%	99.2%	100%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FBP1	95.5%	94.8%	100%	100%	99.7%	Fructose-1,6-bisphosphatase deficiency, 229700
FBP2	100%	100%	100%	100%	99.7%	?Leukodystrophy, childhood-onset, remitting, 619864
FBRSL1	100%	99.1%	99.6%	96.9%	89.1%	
FBXL3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	95.6%	95.6%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100%	100%	100%	99.9%	99.3%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO22	100%	100%	100%	100%	99.6%	Tayoun-Maawali syndrome, 621184

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FBXO28	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 100, 619777
FBXO31	100%	100%	100%	100%	99.2%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXO32	100%	100%	100%	100%	99.6%	
FBXO38	100%	100%	100%	100%	99.9%	Neuronopathy, distal hereditary motor, autosomal dominant 6, 615575
FBXO43	100%	100%	100%	100%	99.9%	Spermatogenic failure 64, 619696;Oocyte/zygote/embryo maturation arrest 12, 619697
FBXO7	99.1%	98.1%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FBXW11	100%	100%	100%	100%	99.4%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW4	92.3%	92.3%	100%	100%	99.1%	
FBXW7	100%	100%	100%	100%	99.6%	Developmental delay, hypotonia, and impaired language, 620012
FCGR3A	91.7%	89%	100%	99.9%	99.6%	Immunodeficiency 20, 615707
FCGR3B	99.3%	97.9%	89%	85.4%	74.7%	
FCHO1	98.6%	96.4%	100%	99.9%	99.2%	Immunodeficiency 76, 619164
FCN3	89.9%	89.9%	100%	99.9%	98.8%	Immunodeficiency due to ficolin 3 deficiency, 613860
FCSK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation with defective fucosylation 2, 618324

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FDFT1	100%	98.7%	100%	100%	99.5%	Squalene synthase deficiency, 618156
FDPS	100%	100%	100%	100%	99.3%	Porokeratosis 9, multiple types, 616631
FDX2	100%	100%	100%	100%	99.3%	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy, 251900
FDXR	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 9B, 620887;Auditory neuropathy and optic atrophy, 617717
FECH	100%	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FEM1B	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities, 621263
FERMT1	100%	100%	100%	100%	99.7%	Kindler syndrome, 173650
FERMT3	100%	100%	100%	100%	99.4%	Leukocyte adhesion deficiency, type III, 612840
FERRY3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 66, 618221
FEZF1	100%	100%	100%	99.9%	99.6%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FEZF2	100%	100%	100%	100%	98.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGA	100%	99.7%	100%	100%	99.7%	Amyloidosis, hereditary systemic 2, 105200;Hypodysfibrinogenemia, congenital, 616004;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGB	100%	100%	100%	100%	99.6%	Hypofibrinogenemia, congenital, 202400;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGD1	100%	100%	98.4%	88%	69.3%	Aarskog-Scott syndrome, 305400
FGD4	100%	100%	100%	100%	99.8%	Charcot-Marie-Tooth disease, type 4H, 609311
FGF10	100%	100%	100%	100%	99.9%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF12	100%	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 47, 617166
FGF13	100%	99.6%	99.6%	91.4%	73.2%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095
FGF14	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174
FGF16	100%	100%	98.4%	87.9%	69%	Metacarpal 4-5 fusion, 309630
FGF17	100%	100%	100%	100%	99.5%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGF20	100%	100%	100%	100%	99.6%	?Renal hypodysplasia/aplasia 2, 615721
FGF23	100%	100%	100%	100%	99.2%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	100%	100%	100%	99.8%	97.8%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF4	100%	100%	100%	99.9%	98.4%	Short-rib thoracic dysplasia 22 without polydactyly, 621260
FGF5	100%	100%	100%	100%	99.6%	Trichomegaly, 190330
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	100%	99.8%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	99.2%	100%	100%	99.4%	Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGFR2	100%	100%	100%	100%	99.7%	Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGFR3	100%	100%	100%	99.9%	99.1%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FGG	100%	100%	100%	100%	99.8%	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, 616004;Hypofibrinogenemia, congenital, 202400;Afibrinogenemia, congenital, 202400
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FHL1	100%	100%	99.4%	91.2%	73.3%	Myopathy, X-linked, with postural muscle atrophy, 300696;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;?Uruguay faciocardiomusculoskeletal syndrome, 300280;Scapuloperoneal myopathy, X-linked dominant, 300695;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FHL2	91.7%	91.7%	100%	100%	99.7%	
FHOD3	97.6%	97.6%	100%	100%	99.5%	Cardiomyopathy, familial hypertrophic, 28, 619402
FIBP	98.7%	95%	100%	100%	99.5%	Thauvin-Robinet-Faivre syndrome, 617107
FICD	100%	100%	100%	100%	99.2%	Spastic paraplegia 92, autosomal recessive, 620911
FIG4	98.4%	98.4%	100%	100%	99.9%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FIGLA	100%	100%	100%	100%	99.3%	Premature ovarian failure 6, 612310
FIGN	100%	100%	100%	100%	99.6%	
FIGNL1	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FILIP1	100%	100%	100%	100%	99.7%	Neuromuscular disorder, congenital, with dysmorphic facies, 620775
FITM2	100%	100%	100%	100%	99.2%	Siddiqi syndrome, 618635
FKBP10	100%	100%	100%	100%	99.3%	Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKBP14	100%	100%	100%	100%	99.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FKBP6	100%	100%	100%	99.9%	99.6%	Spermatogenic failure 77, 620103
FKRP	100%	100%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FKTN	95.2%	94.9%	100%	100%	99.9%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152
FLAD1	100%	100%	100%	100%	99.5%	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100
FLCN	96.9%	96.9%	100%	100%	99.7%	Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700
FLG	100%	100%	99.8%	99.7%	99.1%	Ichthyosis vulgaris, 146700;{Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	100%	100%	99.9%	99.9%	99.5%	Peeling skin syndrome 6, 618084
FLI1	91.9%	91.9%	100%	100%	99.5%	Bleeding disorder, platelet-type, 21, 617443
FLII	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 2J, 620635

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FLNA	100%	100%	98.3%	87.4%	68.7%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FLNB	100%	100%	100%	100%	99.6%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310
FLNC	100%	100%	100%	100%	99.6%	Cardiomyopathy, familial dilated, 1PP, 617047;Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
FLRT3	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 21 with anosmia, 615271

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FLT3	100%	100%	100%	100%	99.8%	Leukemia, acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, reduced survival in, somatic, 601626;Leukemia, acute myeloid, somatic, 601626
FLT3LG	100%	100%	100%	99.9%	98.7%	?Immunodeficiency 125, 620926
FLT4	99.1%	97.6%	100%	100%	99.4%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelop mental disorder with microcephaly, absent speech, and hypotonia, 621060
FLVCR2	100%	100%	100%	100%	99.7%	Proliferative vasculopathy and hydranencephaly-hydro cephaly syndrome, 225790
FMN1	100%	100%	100%	100%	99.6%	
FMN2	100%	100%	99.9%	99.7%	97.9%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMO3	100%	100%	100%	100%	99.9%	Trimethylaminuria, 602079
FMR1	97%	97%	99.2%	92.8%	75.4%	Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FN1	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia, corner fracture type, 184255;Glomerulopathy with fibronectin deposits 2, 601894
FNIP1	100%	100%	100%	99.9%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOCAD	100%	100%	100%	99.9%	99.7%	Liver disease, severe congenital, 619991
FOLR1	100%	100%	100%	100%	99.5%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOSL2	100%	100%	100%	99.9%	99.2%	Aplasia cutis-enamel dysplasia syndrome, 620789
FOXC1	100%	100%	100%	99.4%	94.5%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXC2	100%	100%	100%	99.8%	98.2%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	100%	100%	100%	99.9%	97.1%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534
FOXE3	100%	100%	100%	99.8%	97.4%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FOXF1	100%	100%	99.9%	99.2%	96.2%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FOXF2	99.9%	99.4%	100%	99.7%	97.4%	
FOXG1	100%	100%	100%	99.9%	98.2%	Rett syndrome, congenital variant, 613454
FOXH1	100%	100%	100%	100%	99.5%	
FOXI1	100%	100%	100%	100%	99.8%	Enlarged vestibular aqueduct, 600791
FOXI3	100%	99.9%	100%	99.5%	97.6%	Craniofacial microsomia 2, 620444
FOXJ1	100%	100%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 43, 618699
FOXL1	100%	100%	100%	99.9%	98.1%	Otosclerosis 11, 620576
FOXL2	100%	100%	99.9%	99.4%	96.8%	Premature ovarian failure 3, 608996;Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100
FOXN1	100%	100%	100%	100%	99.8%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705;?T-cell immunodeficiency with thymic aplasia, 242700
FOXO1	100%	100%	100%	99.7%	97.7%	Rhabdomyosarcoma, alveolar, 268220
FOXP1	100%	100%	100%	100%	99.9%	Intellectual developmental disorder with language impairment with or without autistic features, 613670

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FOXP2	100%	100%	100%	100%	99.7%	Speech-language disorder-1, 602081
FOXP3	100%	100%	99.1%	88.7%	71.1%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FOXP4	100%	100%	100%	100%	99.3%	
FOXRED1	100%	100%	100%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FPR1	100%	100%	100%	100%	99.3%	
FRA10AC1	92.2%	92.2%	100%	99.9%	99.5%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113
FRAS1	100%	100%	100%	100%	99.7%	Fraser syndrome 1, 219000
FREM1	100%	100%	100%	100%	99.6%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FREM2	100%	100%	100%	100%	99.7%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FRMD4A	96.6%	96.6%	100%	99.9%	99.5%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD5	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FRMD7	100%	100%	99.5%	93.2%	75.5%	Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700
FRMPD4	100%	100%	98.7%	88.4%	70.1%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100%	100%	100%	100%	98.2%	Developmental and epileptic encephalopathy 37, 616981
FRYL	100%	100%	100%	99.9%	99.6%	Pan-Chung-Bellen syndrome, 621049
FSCN2	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 30, 607921
FSD1L	100%	100%	100%	99.9%	99.6%	
FSHB	100%	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	100%	99.8%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FSIP2	100%	100%	100%	99.9%	99.5%	Spermatogenic failure 34, 618153
FTCD	100%	100%	100%	99.8%	98.7%	Glutamate formiminotransferase deficiency, 229100
FTH1	100%	100%	100%	100%	99.7%	Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FTL	100%	100%	100%	100%	99.3%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
FTO	94.5%	94.5%	100%	100%	99.9%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	100%	100%	98.6%	86.8%	71.1%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100%	100%	100%	100%	99.5%	Fucosidosis, 230000
FURIN	100%	99.1%	100%	100%	99.7%	
FUS	100%	100%	100%	100%	99.2%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030;Essential tremor, hereditary, 4, 614782
FUT2	100%	100%	100%	99.8%	98.4%	{Norwalk virus infection, resistance to};{Vitamin B12 plasma level QTL1}, 612542;[Bombay phenotype, digenic], 616754
FUT6	100%	100%	100%	100%	99.5%	[Fucosyltransferase 6 deficiency], 613852
FUT8	97.2%	97.2%	100%	100%	99.8%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FUZ	93.7%	93.7%	100%	100%	99.2%	{Neural tube defects, susceptibility to}, 182940

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FXN	100%	100%	100%	100%	98.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
FXR1	100%	100%	100%	99.9%	99.4%	Congenital myopathy 9B, proximal, with minicore lesions, 618823;?Congenital myopathy 9A with respiratory insufficiency and bone fractures, 618822
FXYD2	80.6%	80.4%	100%	99.7%	99%	Hypomagnesemia 2, renal, 154020
FYB1	100%	100%	100%	100%	99.9%	Thrombocytopenia 3, 273900
FYCO1	100%	100%	100%	100%	99.5%	Cataract 18, autosomal recessive, 610019
FZD2	100%	100%	100%	99.9%	98.6%	Omodysplasia 2, 164745
FZD4	100%	100%	100%	99.8%	99%	Retinopathy of prematurity, 133780;Exudative vitreoretinopathy 1, 133780
FZD5	100%	100%	100%	100%	98.9%	Microphthalmia/colobo ma 11, 620731
FZD6	99.9%	99.1%	100%	100%	99.8%	Nail disorder, nonsyndromic congenital, 1, 161050
FZR1	99.7%	97.1%	100%	100%	99.5%	Developmental and epileptic encephalopathy 109, 620145
G6PC1	100%	100%	100%	100%	99.7%	Glycogen storage disease Ia, 232200
G6PC3	96.8%	96.8%	100%	100%	99.8%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

G6PD	86.3%	86.3%	99%	89.1%	71.2%	Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient, 300908;{Resistance to malaria due to G6PD deficiency}, 611162
GAA	100%	100%	100%	100%	99.5%	Pompe disease, late-onset, 621314;Pompe disease, infantile-onset, 232300
GAB1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 26, 605428
GABBR1	100%	100%	100%	99.9%	98.9%	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502
GABBR2	100%	100%	100%	100%	99.4%	{Nicotine dependence, protection against}, 188890;{Nicotine dependence, susceptibility to}, 188890;Developmental and epileptic encephalopathy 59, 617904;Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	100%	100%	100%	100%	99.4%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRA2	95%	95%	100%	100%	99.8%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GABRA3	100%	100%	98.7%	90.1%	71%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRA4	100%	100%	100%	100%	99.9%	
GABRA5	100%	100%	100%	99.9%	99.5%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100%	100%	100%	100%	99.7%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113
GABRD	100%	100%	100%	99.7%	98.3%	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060
GABRG2	92.9%	92.9%	100%	100%	99.7%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 89, 619124

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GAL	70.3%	69.6%	100%	100%	99.5%	?Epilepsy, familial temporal lobe, 8, 616461
GALC	100%	100%	100%	100%	99.6%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	99.6%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALK1	92.3%	92.3%	100%	100%	99.8%	Galactokinase deficiency with cataracts, 230200
GALM	84%	84%	100%	100%	99.7%	Galactosemia IV, 618881
GALNS	100%	100%	100%	100%	99%	Mucopolysaccharidosis IVA, 253000
GALNT12	90.6%	90.6%	100%	100%	99.4%	{Colorectal cancer, susceptibility to, 1}, 608812
GALNT14	100%	100%	100%	99.9%	99.4%	
GALNT2	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIt, 618885
GALNT3	100%	100%	100%	100%	99.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GALNTL5	100%	100%	100%	100%	99.7%	
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400
GAMT	97%	93.2%	100%	99.8%	98.8%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	100%	100%	99.9%	99.5%	Giant axonal neuropathy-1, 256850
GANAB	100%	100%	100%	100%	99.6%	Polycystic kidney disease 3, 600666
GAPVD1	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GARS1	98.9%	98.9%	100%	100%	99.8%	Spinal muscular atrophy, infantile, James type, 619042;Neuronopathy, distal hereditary motor, autosomal dominant 5, 600794;Charcot-Marie-Tooth disease, type 2D, 601472
GAS2	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 125, 620877
GAS2L2	100%	100%	100%	100%	99.4%	?Ciliary dyskinesia, primary, 41, 618449
GAS8	100%	100%	100%	99.9%	99.6%	
GATA1	100%	100%	98.9%	88.4%	68.2%	Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367;Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835;Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	85.7%	85.7%	100%	100%	99.1%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GATA3	100%	100%	100%	100%	98.6%	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GATA4	100%	100%	100%	99.9%	98.2%	Tetralogy of Fallot, 187500;Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542
GATA5	100%	99.1%	100%	99.9%	98.6%	Congenital heart defects, multiple types, 5, 617912
GATA6	93.8%	93.8%	100%	99.8%	98.1%	Atrial septal defect 9, 614475;Persistent truncus arteriosus, 217095;Pancreatic agenesis and congenital heart defects, 600001;Atrioventricular septal defect 5, 614474;Tetralogy of Fallot, 187500
GATAD1	100%	100%	100%	99.9%	99.2%	?Cardiomyopathy, dilated, 2B, 614672
GATAD2A	100%	100%	100%	99.9%	99.5%	
GATAD2B	100%	100%	100%	100%	99.4%	GAND syndrome, 615074
GATB	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 41, 618838
GATC	80.1%	80.1%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 42, 618839
GATM	100%	100%	100%	100%	99.9%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	100%	100%	100%	99.9%	99.4%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GBF1	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2GG, 606483
GCDH	79.8%	79.8%	100%	99.9%	99.5%	Glutaricaciduria, type I, 231670
GCGR	100%	100%	100%	99.9%	99.4%	Mahvash disease, 619290
GCH1	100%	100%	100%	100%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GCK	100%	100%	100%	99.9%	99.2%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GCLC	100%	100%	100%	100%	99.6%	{Myocardial infarction, susceptibility to}, 608446;Anemia, congenital, nonspherocytic hemolytic, 7, 230450
GCLM	93.1%	93.1%	100%	100%	100%	{Myocardial infarction, susceptibility to}, 608446
GCM2	100%	100%	100%	100%	99.7%	Hypoparathyroidism, familial isolated 2, 618883;Hyperparathyroidism 4, 617343
GCNA	100%	100%	98.8%	89.9%	71.3%	Spermatogenic failure, X-linked, 4, 301077
GCNT2	100%	100%	100%	100%	99.6%	[Blood group, Ii], 110800;Adult i phenotype without cataract, 110800;Cataract 13 with adult i phenotype, 116700
GCSH	100%	100%	100%	100%	99.9%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDAP1	86.2%	86.2%	98.3%	98%	97.1%	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706;Charcot-Marie-Tooth disease, recessive intermediate, A, 608340;Charcot-Marie-Tooth disease, axonal, type 2K, 607831;Charcot-Marie-Tooth disease, type 4A, 214400
GDAP2	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GDF1	100%	100%	100%	100%	98.9%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GDF11	100%	100%	100%	99.6%	98.6%	?Vertebral hypersegmentation and orofacial anomalies, 619122
GDF2	100%	100%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF3	100%	100%	100%	100%	99.6%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704
GDF5	100%	100%	100%	100%	99.2%	Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072
GDF6	100%	100%	100%	99.9%	98.2%	Microphthalmia with coloboma 6, digenic, 613703;Microphthalmia, isolated 4, 613094;Leber congenital amaurosis 17, 615360;Multiple synostoses syndrome 4, 617898;Klippel-Feil syndrome 1, autosomal dominant, 118100
GDF9	100%	100%	100%	100%	99.9%	Premature ovarian failure 14, 618014

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GDI1	100%	100%	99%	88.1%	69.4%	Intellectual developmental disorder, X-linked 41, 300849
GDNF	100%	100%	100%	100%	99.3%	{Hirschsprung disease, susceptibility to, 3}, 613711
GDPD1	100%	100%	100%	100%	99.8%	
GEMIN4	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	100%	100%	100%	99.9%	99.6%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GET3	87.9%	87.9%	100%	99.9%	98.9%	?Cardiomyopathy, dilated, 2H, 620203
GET4	100%	100%	100%	100%	99.5%	?Congenital disorder of glycosylation, type IIy, 620200
GFAP	100%	100%	100%	99.9%	99.4%	Alexander disease, 203450
GFER	100%	100%	100%	99.9%	99.4%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFI1	100%	100%	100%	100%	99.2%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GFI1B	100%	100%	100%	100%	99.3%	Bleeding disorder, platelet-type, 17, 187900
GFM1	96.6%	96.6%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 1, 609060

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GFM2	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 39, 618397
GFPT1	97.5%	97.5%	100%	100%	99.8%	Myasthenia, congenital, 12, with tubular aggregates, 610542
GFRA1	94%	94%	100%	100%	99.5%	Renal hypodysplasia/aplasia 4, 619887
GFUS	100%	100%	100%	100%	99.5%	
GGCX	100%	100%	100%	100%	99.4%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GGN	100%	100%	100%	99.9%	98.6%	Spermatogenic failure 69, 619826
GGPS1	100%	100%	100%	100%	99.9%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GGT1	100%	100%	100%	99.9%	99.4%	?Glutathioninuria, 231950
GH1	100%	100%	100%	100%	99.5%	Kowarski syndrome, 262650;Growth hormone deficiency, isolated, type II, 173100;Growth hormone deficiency, isolated, type IB, 612781;Growth hormone deficiency, isolated, type IA, 262400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GHR	99.6%	99.6%	99.8%	99.8%	99.1%	Laron dwarfism, 262500;Increased responsiveness to growth hormone, 604271;Growth hormone insensitivity, partial, 604271;{Hypercholesterolemia, familial, modifier of}, 143890
GHRHR	100%	100%	100%	100%	99.4%	Growth hormone deficiency, isolated, type IV, 618157
GHSR	100%	100%	100%	99.9%	99.4%	Growth hormone deficiency, isolated partial, 615925
GIGYF1	100%	100%	100%	100%	99%	
GIMAP5	100%	100%	100%	100%	99.9%	Portal hypertension, noncirrhotic, 2, 619463
GIMAP6	100%	100%	100%	100%	99.7%	
GINS1	81%	81%	100%	100%	99.8%	Immunodeficiency 55, 617827
GINS2	100%	100%	100%	99.9%	99.5%	
GINS3	100%	99.9%	100%	100%	99.6%	Meier-Gorlin syndrome 9, 621512
GINS4	100%	100%	100%	100%	99.9%	
GIPC1	100%	100%	100%	99.8%	99.1%	Oculopharyngodistal myopathy 2, 618940
GIPC3	100%	100%	100%	100%	99%	Deafness, autosomal recessive 15, 601869

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GJA1	100%	100%	100%	100%	99.7%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GJA3	100%	100%	100%	99.9%	98.8%	Cataract 14, multiple types, 601885
GJA5	100%	100%	100%	100%	99.9%	Atrial fibrillation, familial, 11, 614049;Atrial standstill, digenic (GJA5/SCN5A), 108770
GJA8	100%	100%	100%	100%	99.6%	Cataract 1, multiple types, 116200
GJB1	100%	100%	97.9%	84.9%	64.7%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJB2	100%	100%	100%	100%	99.6%	Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GJB3	100%	100%	100%	100%	99.5%	Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB4	100%	100%	100%	100%	99.9%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100%	100%	100%	99.9%	99.8%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290
GJC2	100%	99.4%	100%	99.9%	98.3%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GK	96%	96%	99.5%	92.1%	73.6%	Glycerol kinase deficiency, 307030
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
GLB1	100%	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	97.3%	97.3%	100%	100%	99.6%	Glycine encephalopathy1, 605899

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GLDN	100%	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 11, 617194
GLE1	100%	100%	100%	100%	99.7%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890
GLI1	100%	100%	100%	99.9%	99.1%	Polydactyly, preaxial I, 174400;Polydactyly, postaxial, type A8, 618123
GLI2	100%	100%	100%	99.9%	99.3%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GLI3	99.3%	99.3%	100%	99.9%	99.2%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GLIS1	100%	100%	100%	100%	99%	
GLIS2	100%	100%	100%	99.8%	99.1%	Nephronophthisis 7, 611498
GLIS3	100%	100%	100%	100%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLIS3-AS1						
GLMN	96.2%	96.2%	100%	100%	99.9%	Glomuvenous malformations, 138000
GLRA1	100%	100%	100%	100%	99.7%	Hyperekplexia 1, 149400
GLRA2	100%	100%	99.2%	91.6%	73.5%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GLRB	93.7%	93.7%	100%	100%	99.7%	Hyperekplexia 2, 614619
GLRX5	100%	100%	100%	100%	99.7%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859
GLS	95.8%	95.8%	100%	100%	99.8%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100%	100%	100%	100%	99.8%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100%	100%	100%	99.9%	99.2%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806
GLYCTK	100%	100%	100%	99.9%	99.3%	D-glyceric aciduria, 220120
GLYR1	100%	100%	100%	100%	99.2%	
GM2A	100%	100%	100%	100%	99.7%	GM2-gangliosidosis, AB variant, 272750
GMNN	91.4%	91.4%	100%	100%	99.8%	Meier-Gorlin syndrome 6, 616835
GMPPA	100%	100%	100%	100%	98.8%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GMPPB	100%	100%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
GMPR	87.3%	87.3%	100%	100%	99.6%	
GMPS	100%	100%	100%	100%	99.9%	
GNA11	100%	100%	100%	100%	99.5%	Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361
GNA14	100%	100%	100%	100%	99.9%	
GNAI1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854
GNAI2	100%	100%	100%	100%	99.1%	Pituitary adenoma, ACTH-secreting, somatic;Ventricular tachycardia, somatic, 192605
GNAI3	100%	100%	100%	100%	99.9%	Auriculocondylar syndrome 1, 602483
GNAL	100%	100%	100%	99.9%	99.3%	Dystonia 25, 615073
GNAO1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GNAQ	92.8%	92.8%	100%	100%	99.5%	Capillary malformations, congenital, 1, somatic, mosaic, 163000;Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	100%	99.7%	100%	99.9%	98.5%	Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080
GNAS-AS1						Pseudohypoparathyroidism Ib, 603233
GNAT1	100%	100%	100%	100%	98.9%	Night blindness, congenital stationary, autosomal dominant 3, 610444;Night blindness, congenital stationary, type 1G, 616389
GNAT2	100%	100%	100%	100%	99.8%	Achromatopsia 4, 613856
GNB1	94.3%	94.3%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GNB2	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNB3	100%	100%	100%	100%	99.1%	Night blindness, congenital stationary, type 1H, 617024;{Hypertension, essential, susceptibility to}, 145500
GNB4	100%	100%	100%	100%	100%	Charcot-Marie-Tooth disease, dominant intermediate F, 615185
GNB5	100%	100%	100%	100%	99.8%	Lodder-Merla syndrome, type 2, with developmental delay and with or without cardiac arrhythmia, 617182;Lodder-Merla syndrome, type 1, with impaired intellectual development and cardiac arrhythmia, 617173
GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820
GNMT	100%	100%	100%	99.9%	98.2%	Glycine N-methyltransferase deficiency, 606664
GNPAT	100%	100%	100%	100%	100%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPNAT1	100%	100%	100%	99.9%	99.3%	?Rhizomelic dysplasia, Ain-Naz type, 619598
GNPTAB	100%	100%	100%	100%	99.8%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	90.1%	90.1%	100%	99.8%	98.5%	Mucolipidosis III gamma, 252605

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GNRH1	100%	100%	100%	100%	99.8%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GNS	94.3%	94.3%	100%	100%	99.6%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100%	100%	100%	99.9%	99.1%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GON4L	100%	100%	100%	100%	99.7%	Li-Takada-Miyake syndrome, 621212
GON7	100%	100%	100%	100%	100%	Galloway-Mowat syndrome 9, 619603
GORAB	100%	100%	100%	100%	99.5%	Geroderma osteodysplasticum, 231070
GOSR2	100%	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GOT1	100%	100%	100%	100%	100%	Aspartate aminotransferase, serum level of, QTL1, 614419
GOT2	92.8%	92.8%	100%	100%	99.5%	Developmental and epileptic encephalopathy 82, 618721

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GP1BA	100%	100%	99.9%	99.7%	98.6%	Bernard-Soulier syndrome, type A1 (recessive), 231200;Bernard-Soulier syndrome, type A2 (dominant), 153670;von Willebrand disease, platelet-type, 177820;{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	100%	100%	100%	99.4%	97.5%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GP6	100%	98.2%	100%	100%	99.6%	Bleeding disorder, platelet-type, 11, 614201
GP9	100%	100%	100%	100%	99%	Bernard-Soulier syndrome, type C, 231200
GPAA1	88.5%	88.5%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPAT2	99.6%	99.5%	99.6%	99.2%	97.7%	
GPATCH11	100%	100%	100%	100%	99.8%	
GPC3	94%	94%	98.6%	88.7%	70.8%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100%	100%	99.4%	91.2%	70.7%	Keipert syndrome, 301026
GPC6	100%	100%	100%	100%	99.7%	Omodysplasia 1, 258315
GPD1	100%	100%	100%	100%	99.7%	Hypertriglyceridemia, transient infantile, 614480
GPD1L	100%	100%	100%	100%	99.4%	Brugada syndrome 2, 611777

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GPHN	95.9%	95.9%	100%	100%	99.8%	Molybdenum cofactor deficiency C, 615501
GPI	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 4, glucose phosphate isomerase deficient, 613470
GPIHBP1	94.6%	90.9%	100%	100%	99.9%	Hyperlipoproteinemia, type 1D, 615947
GPNMB	95.1%	95.1%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR101	100%	100%	98.4%	88.9%	67.5%	Pituitary adenoma 2, GH-secreting, 300943
GPR143	93.1%	93.1%	98.6%	89.3%	71.2%	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814
GPR156	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 121, 620551
GPR161	100%	100%	100%	100%	99.5%	{Medulloblastoma predisposition syndrome}, 155255
GPR179	100%	100%	100%	100%	99.5%	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565
GPR68	100%	100%	100%	99.9%	99%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GPR88	100%	100%	100%	99.9%	98.1%	?Chorea, childhood-onset, with psychomotor retardation, 616939
GPRASP2	100%	100%	98.8%	89.2%	72.5%	?Deafness, X-linked 7, 301018
GPRC5B	100%	100%	100%	100%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GPSM2	95.5%	95.5%	100%	100%	99.8%	Chudley-McCullough syndrome, 604213
GPT2	92.9%	92.9%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GPX1	100%	100%	100%	100%	98.9%	Hemolytic anemia due to glutathione peroxidase deficiency, 614164
GPX4	100%	100%	100%	99.9%	98.7%	Spondylometaphyseal dysplasia, Sedaghatian type, 250220
GRAP	100%	100%	100%	99.7%	99.5%	Deafness, autosomal recessive 114, 618456
GREB1L	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GREM1	100%	100%	100%	100%	99.7%	
GREM2	100%	100%	100%	100%	99.2%	Tooth agenesis, selective, 9, 617275
GRHL2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRHL3	100%	100%	100%	100%	99.7%	van der Woude syndrome 2, 606713
GRHPR	86.7%	86.6%	100%	100%	99.7%	Hyperoxaluria, primary, type II, 260000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GRIA1	100%	100%	100%	100%	99.7%	?Intellectual developmental disorder, autosomal recessive 76, 619931;Intellectual developmental disorder, autosomal dominant 67, 619927
GRIA2	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	100%	100%	99.2%	91.4%	73.7%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699
GRIA4	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	95.3%	95.3%	100%	99.9%	99.3%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092
GRIN1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GRIN2A	100%	100%	100%	100%	99.5%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	100%	99.9%	100%	99.8%	97.9%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100%	100%	100%	100%	99.9%	Fraser syndrome 3, 617667
GRK1	100%	100%	100%	100%	98.7%	Oguchi disease-2, 613411
GRM1	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRM6	100%	100%	100%	99.9%	99.2%	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270
GRM7	100%	100%	100%	99.8%	98.4%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100%	100%	100%	100%	99.7%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
GRWD1	100%	100%	100%	100%	99.2%	?Diarrhea 14, congenital, 621160

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GRXCR1	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 25, 613285
GRXCR2	100%	100%	100%	100%	99.6%	?Deafness, autosomal recessive 101, 615837
GSC	100%	100%	100%	99.9%	98.6%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
GSDME	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 5, 600994
GSE1	100%	100%	100%	99.9%	99.2%	
GSK3B	100%	100%	100%	100%	99.8%	
GSN	92.1%	90%	100%	99.8%	98.9%	Amyloidosis, Finnish type, 105120
GSPT2	100%	100%	98.9%	90.3%	74.5%	
GSR	100%	100%	100%	100%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 10, glutathione reductase deficient, 618660
GSS	100%	100%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900;Glutathione synthetase deficiency, 266130
GSX2	100%	100%	100%	100%	99.4%	Diencephalic-mesence phalic junction dysplasia syndrome 2, 618646
GTF2E2	78.7%	78.7%	100%	100%	99.9%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	44.2%	44.2%	100%	100%	99.8%	Trichothiodystrophy 3, photosensitive, 616395
GTF3C3	98.6%	98.5%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies, brain anomalies, and seizures, 621201

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GTF3C5	100%	100%	100%	99.9%	99.4%	
GTPBP1	99.9%	98.3%	100%	99.9%	98.8%	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888
GTPBP2	100%	100%	100%	100%	99.9%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 23, 616198
GUCA1A	100%	100%	100%	100%	98.7%	Cone-rod dystrophy 14, 602093;Cone dystrophy-3, 602093
GUCA1B	100%	100%	100%	99.8%	98.5%	Retinitis pigmentosa 48, 613827
GUCY1A1	100%	100%	100%	100%	99.8%	Moyamoya 6 with achalasia, 615750
GUCY2C	98.7%	98.7%	100%	100%	99.8%	Diarrhea 6, 614616;Meconium ileus, 614665
GUCY2D	100%	100%	100%	100%	99.5%	Cone-rod dystrophy 6, 601777;?Choroidal dystrophy, central areolar 1, 215500;Leber congenital amaurosis 1, 204000;Night blindness, congenital stationary, type 1I, 618555
GUF1	100%	100%	100%	100%	99.6%	?Developmental and epileptic encephalopathy 40, 617065
GUK1	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 21, 621071
GULOP						Scurvy
GUSB	98.4%	98.4%	100%	100%	99.1%	Mucopolysaccharidosis VII, 253220

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GYG1	93.7%	93.7%	100%	100%	100%	?Glycogen storage disease XV, 613507;Polyglucosan body myopathy 2, 616199
GYS1	100%	100%	100%	100%	99.5%	Glycogen storage disease 0, muscle, 611556
GYS2	100%	100%	100%	100%	99.8%	Glycogen storage disease 0, liver, 240600
GZF1	99.3%	97.6%	100%	100%	99.7%	Joint laxity, short stature, and myopia, 617662
H1-4	100%	100%	100%	100%	99.3%	Rahman syndrome, 617537
H19						
H3-3A	100%	100%	100%	99.8%	98.9%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720
H3-3B	100%	100%	100%	100%	99.1%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721
H4C11	100%	100%	100%	99.9%	98.1%	?Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 2, 619759
H4C3	100%	100%	100%	99.8%	98.7%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758
H4C5	100%	100%	100%	100%	98.3%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950
H4C9	100%	100%	100%	99.9%	98.6%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951
H6PD	100%	100%	100%	100%	99.5%	Cortisone reductase deficiency 1, 604931
HAAO	100%	98.9%	100%	100%	99.5%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HABP2	100%	100%	100%	100%	99.9%	{?Thyroid cancer, nonmedullary, 5}, 616535;{Venous thromboembolism, susceptibility to}, 188050
HACD1	71.3%	71.3%	100%	100%	99.6%	Congenital myopathy 11, 619967
HACE1	95.9%	95.9%	100%	100%	99.7%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	87.7%	87.7%	100%	100%	99.6%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	96.1%	95.3%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.7%	Mitochondrial trifunctional protein deficiency 2, 620300
HAGH	100%	100%	100%	99.9%	99.4%	[Glyoxalase II deficiency], 614033
HAMP	100%	100%	100%	100%	99.9%	Hemochromatosis, type 2B, 613313
HAND1	100%	100%	100%	100%	98.9%	
HAND2	100%	100%	100%	99.4%	94%	
HAPLN1	100%	100%	100%	100%	99.9%	
HARS1	93.2%	93.2%	100%	99.9%	99.9%	Charcot-Marie-Tooth disease, axonal, type 2W, 616625;Usher syndrome type 3B, 614504

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HARS2	100%	100%	100%	99.9%	98.9%	Perrault syndrome 2, 614926
HAUS6	92.1%	92.1%	100%	100%	99.9%	
HAVCR2	100%	100%	100%	100%	99.3%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	99.7%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HBA1	100%	100%	100%	100%	99.5%	Hemoglobin H disease, nondeletional, 613978;Thalassemias, alpha-, 604131;Heinz body anemias, alpha-, 140700;Methemoglobinemia, alpha type, 617973;Erythrocytosis, familial, 7, 617981
HBA2	100%	100%	98.3%	94.6%	88.1%	Heinz body anemia, 140700;Thalassemia, alpha-, 604131;Erythrocytosis, familial, 7, 617981;Hemoglobin H disease, deletional and nondeletional, 613978
HBB	100%	100%	100%	100%	99.9%	Methemoglobinemia, beta type, 617971;Thalassemia-beta, dominant inclusion-body, 603902;Sickle cell disease, 603903;Thalassemia, beta, 613985;Delta-beta thalassemia, 141749;{Malaria, resistance to}, 611162;Hereditary persistence of fetal hemoglobin, 141749;Erythrocytosis, familial, 6, 617980;Heinz body anemia, 140700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HBD	100%	100%	100%	100%	99.7%	Thalassemia due to Hb Lepore;Thalassemia, delta-
HBG1	97.3%	96.4%	92.1%	83.5%	71.3%	Fetal hemoglobin quantitative trait locus 1, 141749
HBG2	100%	100%	100%	100%	99.8%	Fetal hemoglobin quantitative trait locus 1, 141749;Cyanosis, transient neonatal, 613977
HCCS	100%	100%	99.3%	91.9%	75.5%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100%	100%	98.5%	88.6%	69.8%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCK	100%	100%	100%	100%	99.4%	Autoinflammation with pulmonary and cutaneous vasculitis, 620296
HCN1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	95.1%	93.5%	99.3%	97.5%	93%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HCN3	100%	100%	100%	99.9%	99.1%	
HCN4	100%	100%	100%	100%	99%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
HCRT	100%	100%	100%	99.7%	98.1%	?Narcolepsy 1, 161400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HDAC3	100%	100%	100%	99.9%	99.3%	
HDAC4	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	94.9%	94.8%	98.7%	89.6%	71.3%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	94.6%	94.6%	99.2%	90.3%	71.3%	Cornelia de Lange syndrome 5, 300882
HDAC9	100%	100%	100%	100%	99.7%	?Auriculocondylar syndrome 4, 620457
HEATR3	100%	100%	100%	100%	99.8%	Diamond-Blackfan anemia 21, 620072
HEATR5B	100%	100%	100%	100%	99.8%	
HECTD1	98.5%	98.5%	100%	100%	99.8%	
HECTD4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250
HECW2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HELLS	97.7%	97.4%	100%	100%	99.8%	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911
HENMT1	92.1%	92.1%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HEPACAM	100%	100%	100%	100%	99.5%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HEPHL1	100%	100%	100%	100%	99.8%	?Abnormal hair, joint laxity, and developmental delay, 261990
HERC1	100%	100%	100%	100%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100%	100%	100%	99.9%	99.5%	Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HES7	100%	100%	100%	99.8%	98.2%	Spondylocostal dysostosis 4, autosomal recessive, 613686
HESX1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100%	100%	100%	100%	99.4%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HEXB	100%	100%	100%	99.9%	99.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HEY2	100%	100%	100%	99.9%	98.8%	
HFE	100%	99.7%	100%	100%	99.4%	Hemochromatosis, type 1, 235200
HFM1	100%	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HGD	90.1%	90%	100%	100%	99.9%	Alkaptonuria, 203500
HGF	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 39, 608265
HGSNAT	83.5%	83.5%	100%	100%	99.8%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HHAT	100%	100%	100%	100%	99.6%	Nivelon-Nivelon-Mabille syndrome, 600092
HIBADH	73.7%	73.7%	100%	100%	99.8%	
HIBCH	100%	100%	100%	100%	100%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983
HIKESHI	100%	100%	100%	100%	99.4%	Leukodystrophy, hypomyelinating, 13, 616881
HINT1	100%	100%	100%	100%	99.8%	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200
HIVEP2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 43, 616977
HJV	100%	100%	100%	100%	99.8%	Hemochromatosis, type 2A, 602390

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelop mental disorder with visual defects and brain anomalies, 618547
HKDC1	100%	100%	100%	99.9%	99.3%	Retinitis pigmentosa 92, 619614
HLCS	100%	100%	100%	100%	99.2%	Holocarboxylase synthetase deficiency, 253270
HLX	100%	100%	100%	100%	99.3%	
HMBS	100%	100%	100%	100%	99.9%	Leukoencephalopathy, porphyria-related, 620711;Encephalopath y, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HMCN1	100%	100%	100%	100%	99.8%	{Macular degeneration, age-related, 1}, 603075
HMGA2	93.9%	85.5%	100%	100%	99.6%	Silver-Russell syndrome 5, 618908
HMGB1	100%	100%	100%	100%	99.4%	
HMGB3	100%	100%	99.1%	89.3%	70.4%	?Microphthalmia, syndromic 13, 300915
HMGCL	76.2%	76.2%	100%	100%	99.5%	HMG-CoA lyase deficiency, 246450

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HMGCR	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 28, 620375;[Statins, response to], 620410;[Low density lipoprotein cholesterol level QTL 3], 620410
HMGCS1	97.2%	97.2%	100%	100%	99.7%	Congenital myopathy 28 with rigid spine, 621433
HMGCS2	97.5%	97.5%	100%	100%	99.2%	HMG-CoA synthase-2 deficiency, 605911
HMOX1	100%	100%	100%	100%	99.5%	Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HMX1	100%	100%	99.9%	99.5%	97.7%	Oculoauricular syndrome, 612109
HNF1A	100%	100%	100%	100%	99.5%	Hepatic adenoma, somatic, 142330;Diabetes mellitus, insulin-dependent, 20, 612520;{Diabetes mellitus, noninsulin-dependent, 2}, 125853;MODY, type III, 600496;{Diabetes mellitus, insulin-dependent}, 222100;Renal cell carcinoma, 144700
HNF1B	100%	100%	100%	100%	99.6%	Type 2 diabetes mellitus, 125853;Renal cysts and diabetes syndrome, 137920;{Renal cell carcinoma}, 144700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HNF4A	100%	100%	100%	100%	99.3%	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026;{Diabetes mellitus, noninsulin-dependent}, 125853;MODY, type I, 125850
HNMT	100%	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807
HNRNPA1	100%	100%	100%	100%	99.7%	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424;?Myopathy, distal, 3, 610099;Amyotrophic lateral sclerosis 20, 615426
HNRNPA2B1	100%	100%	100%	100%	99.7%	Oculopharyngeal muscular dystrophy 2, 620460;?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422
HNRNPC	87.6%	87.6%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 74, 620688
HNRNPD	100%	100%	100%	100%	99.9%	
HNRNPDL	100%	100%	100%	99.7%	97.5%	Muscular dystrophy, limb-girdle, autosomal dominant 3, 609115

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HNRNPH1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083
HNRNPH2	100%	100%	99.4%	91.3%	73.6%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986
HNRNPK	98.7%	96%	100%	100%	99.7%	Au-Kline syndrome, 616580
HNRNPR	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HNRNPU	98.5%	98.5%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 54, 617391
HOGA1	100%	100%	100%	100%	99.5%	Hyperoxaluria, primary, type III, 613616
HOMER2	93.8%	93.8%	100%	100%	99.8%	?Deafness, autosomal dominant 68, 616707
HORMAD2	100%	100%	100%	100%	99.4%	
HOXA1	100%	100%	100%	100%	99.5%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536
HOXA11	100%	100%	100%	100%	99.1%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HOXA13	100%	99.8%	99.6%	96.3%	86%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HOXA2	100%	100%	100%	99.6%	98%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HOXB1	100%	100%	100%	100%	99.9%	Facial paresis, hereditary congenital, 3, 614744
HOXB13	100%	100%	100%	100%	99.4%	{Prostate cancer, hereditary, 9}, 610997
HOXC13	100%	100%	100%	100%	99.4%	Ectodermal dysplasia 9, hair/nail type, 614931
HOXD10	100%	100%	100%	100%	98.6%	Vertical talus, congenital, 192950;Charcot-Marie-Tooth disease, foot deformity of, 192950
HOXD13	100%	100%	100%	99.9%	98.8%	Syndactyly, type V, 186300;Synpolydactyly 1, 186000;Brachydactyly, type E, 113300;Brachydactyly, type D, 113200;?Brachydactyly-syndactyly syndrome, 610713
HPCA	100%	100%	100%	99.9%	98.8%	Dystonia 2, torsion, autosomal recessive, 224500
HPD	93.7%	90.9%	100%	99.9%	98.9%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HPGD	100%	100%	100%	100%	99.9%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100
HPRT1	87.6%	87.4%	99%	91.6%	74%	Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322
HPS1	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	100%	99.4%	Hermansky-Pudlak syndrome 6, 614075
HPSE2	100%	100%	100%	100%	99.5%	Urofacial syndrome 1, 236730
HR	100%	100%	100%	100%	99.5%	Atrichia with papular lesions, 209500;Alopecia universalis, 203655

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HRAS	100%	100%	100%	99.9%	99.5%	Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
HRG	95.1%	93.3%	100%	100%	99.4%	Thrombophilia 11 due to HRG deficiency, 613116
HROB	95.2%	95.2%	100%	100%	99.4%	Ovarian dysgenesis 11, 620897
HRURF	100%	100%	100%	100%	100%	Hypotrichosis 4, 146550
HS2ST1	100%	100%	100%	100%	99.7%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS3ST6	100%	100%	100%	99.9%	97.8%	?Angioedema, hereditary, 8, 619367
HS6ST1	100%	100%	100%	100%	98.6%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HS6ST2	100%	100%	98.8%	87.6%	70%	?Paganini-Miozzo syndrome, 301025
HSCB	85.5%	85.5%	100%	99.8%	99.1%	?Anemia, sideroblastic, 5, 619523
HSD11B1	100%	100%	100%	100%	99.9%	Cortisone reductase deficiency 2, 614662
HSD11B2	100%	100%	100%	100%	99.4%	Apparent mineralocorticoid excess, 218030

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HSD17B10	100%	100%	99.5%	91%	71.8%	HSD10 mitochondrial disease, 300438
HSD17B3	100%	100%	100%	100%	99.8%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	100%	100%	100%	100%	99.6%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSD3B7	100%	100%	100%	100%	99.5%	Bile acid synthesis defect, congenital, 1, 607765
HSF2	100%	100%	100%	100%	99.8%	
HSF2BP	100%	100%	100%	100%	99.9%	Premature ovarian failure 19, 619245
HSF4	100%	100%	100%	99.9%	99.2%	Cataract 5, multiple types, 116800
HSFY1	50%	50%	49.1%	42.7%	26.8%	
HSFY2	50%	50%	49.8%	44.4%	29%	
HSPA9	100%	100%	100%	100%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPB1	100%	100%	100%	100%	99.1%	Charcot-Marie-Tooth disease, axonal, type 2F, 606595;Neuronopathy, distal hereditary motor, autosomal dominant 3, 608634
HSPB3	100%	100%	100%	100%	99.2%	?Neuronopathy, distal hereditary motor, autosomal dominant 4, 613376
HSPB6	100%	100%	100%	99.6%	97.6%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HSPB8	100%	100%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal dominant 2, 158590;Charcot-Marie-Tooth disease, axonal, type 2L, 608673;Myopathy, myofibrillar, 13, with rimmed vacuoles, 621078
HSPD1	99.9%	98.9%	100%	100%	99.9%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HSPG2	100%	100%	100%	100%	99.4%	Dyssegmental dysplasia, Silverman-Handmaker type, 224410;Schwartz-Jampel syndrome, type 1, 255800
HTR1A	100%	100%	100%	99.9%	99.2%	?Periodic fever, menstrual cycle dependent, 614674
HTRA1	100%	100%	100%	99.9%	99.3%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142
HTRA2	100%	100%	100%	100%	99.5%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248
HTT	99.9%	99.9%	100%	100%	99.6%	Lopes-Maciel-Rodan syndrome, 617435;Huntington disease, 143100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

HUWE1	100%	100%	98.9%	90.1%	71.6%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
HYAL1	100%	100%	100%	99.9%	98.9%	Mucopolysaccharidosis type IX, 601492
HYAL2	100%	100%	100%	100%	99.5%	Muggenthaler-Chowdhury-Chioza syndrome, 621063
HYCC1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
HYDIN	100%	99.9%	100%	99.9%	99.5%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	100%	99.8%	Hydrolethalus syndrome, 236680
HYOU1	100%	100%	100%	100%	99.3%	?Immunodeficiency 59 and hypoglycemia, 233600
IARS1	100%	100%	100%	100%	99.6%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	99.7%	100%	99.9%	99.6%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
ICOS	100%	100%	100%	99.9%	99.7%	Immunodeficiency, common variable, 1, 607594
ICOSLG	100%	100%	100%	100%	99.6%	?Immunodeficiency 119, 620825
ID4	100%	100%	100%	99.9%	98.4%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IDH1	100%	100%	100%	99.9%	99.6%	{Glioma, susceptibility to, somatic}, 137800
IDH2	89.7%	89.5%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria 2, 613657
IDH3A	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 90, 619007
IDH3B	100%	100%	100%	100%	99%	Retinitis pigmentosa 46, 612572
IDH3G	100%	100%	98.6%	88%	69.7%	Retinitis pigmentosa 99, 301148
IDI1	100%	100%	100%	100%	99.5%	
IDS	100%	100%	99.4%	91.4%	72.9%	Mucopolysaccharidosis II, 309900
IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IER3IP1	90.2%	74.9%	100%	100%	99.3%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	100%	100%	99.8%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFITM5	100%	100%	100%	100%	99.7%	Osteogenesis imperfecta, type V, 610967
IFNAR1	94.5%	94.5%	100%	100%	99.6%	Immunodeficiency 106, susceptibility to viral infections, 619935
IFNAR2	100%	100%	100%	100%	99.9%	{Hepatitis B virus, susceptibility to}, 610424;Immunodeficiency 45, 616669

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IFNG	100%	100%	100%	100%	100%	{Hepatitis C virus, response to therapy of}, 609532;{TSC2 angiomyolipomas, renal, modifier of}, 613254;{Aplastic anemia}, 609135;?Immunodeficiency 69, mycobacteriosis, 618963;{Tuberculosis, protection against}, 607948;{AIDS, rapid progression to}, 609423
IFNGR1	100%	100%	100%	100%	99.8%	{H. pylori infection, susceptibility to}, 600263;Immunodeficiency 27A, mycobacteriosis, AR, 209950;Immunodeficiency 27B, mycobacteriosis, AD, 615978;{Tuberculosis infection, protection against}, 607948;{Tuberculosis, susceptibility to}, 607948;{Hepatitis B virus infection, susceptibility to}, 610424
IFNGR2	91.6%	91.6%	100%	99.9%	99.4%	Immunodeficiency 28, mycobacteriosis, 614889
IFNLR1	100%	100%	100%	99.9%	99.5%	
IFRD1	100%	100%	100%	100%	99.6%	
IFT122	100%	100%	100%	100%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	99.8%	96.5%	100%	100%	99.9%	Bardet-Biedl syndrome 19, 615996
IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	92.9%	92.9%	100%	100%	99.8%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT56	100%	100%	100%	100%	99.8%	Biliary, renal, neurologic, and skeletal syndrome, 619534
IFT57	92.4%	92.4%	100%	100%	99.9%	?Orofaciodigital syndrome XVIII, 617927
IFT74	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT80	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.5%	92.5%	100%	100%	99.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IFT88	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IGBP1	100%	100%	98.8%	91.9%	72.1%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100%	100%	100%	99.9%	99.1%	Insulin-like growth factor I deficiency, 608747
IGF1R	100%	100%	100%	100%	99.7%	Insulin-like growth factor I, resistance to, 270450
IGF2	100%	100%	100%	100%	99.7%	Silver-Russell syndrome 3, 616489
IGF2R	100%	100%	100%	100%	99.5%	Hepatocellular carcinoma, somatic, 114550
IGFALS	95.7%	95.7%	100%	100%	99.2%	Acid-labile subunit, deficiency of, 615961
IGFBP7	85.5%	85.5%	100%	99.9%	98.8%	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224
IGHG2	100%	100%	99.5%	97.9%	94.7%	
IGHM	100%	100%	100%	100%	99.8%	
IGHMBP2	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
IGKC	100%	100%	100%	100%	99.8%	
IGLL1	100%	100%	100%	100%	99.2%	Agammaglobulinemia 2, 613500
IGSF1	100%	100%	99.3%	91.1%	73.4%	Hypothyroidism, central, and testicular enlargement, 300888
IGSF10	100%	100%	100%	100%	99.8%	
IGSF3	100%	100%	100%	100%	99.6%	?Lacrimal duct defect, 149700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IHH	100%	100%	100%	99.8%	99%	Acrocapitofemoral dysplasia, 607778;Brachydactyly, type A1, 112500
IKBKB	100%	100%	100%	100%	99.4%	Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	94.1%	93.5%	99%	90.5%	73.6%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IKZF1	100%	100%	100%	100%	99.5%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	100%	100%	99.9%	Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233
IKZF3	100%	100%	100%	100%	99.8%	?Immunodeficiency 84, 619437
IKZF5	100%	100%	100%	100%	99.8%	Thrombocytopenia, autosomal dominant, 7, 619130
IL10	100%	100%	100%	100%	99.7%	{Rheumatoid arthritis, progression of}, 180300;{Graft-versus-host disease, protection against}, 614395;{HIV-1, susceptibility to}, 609423
IL10RA	100%	100%	100%	100%	99.6%	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IL10RB	100%	100%	100%	100%	99.7%	{Hepatitis B virus, susceptibility to}, 610424;Inflammatory bowel disease 25, early onset, autosomal recessive, 612567
IL11RA	92.1%	92.1%	100%	100%	99.3%	Craniosynostosis and dental anomalies, 614188
IL12B	100%	100%	100%	100%	100%	Immunodeficiency 29, mycobacteriosis, 614890
IL12RB1	94.1%	94.1%	100%	99.9%	99.2%	Immunodeficiency 30, 614891
IL17F	100%	100%	100%	100%	100%	?Candidiasis, familial, 6, autosomal dominant, 613956
IL17RA	100%	100%	100%	100%	99.5%	Immunodeficiency 51, 613953
IL17RC	100%	100%	100%	99.9%	99.3%	Candidiasis, familial, 9, 616445
IL17RD	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL18BP	100%	100%	100%	99.8%	99.5%	{?Hepatitis, fulminant viral, susceptibility to}, 618549
IL1R1	97.7%	97.7%	100%	100%	99.9%	?Chronic recurrent multifocal osteomyelitis 3, 259680
IL1RAPL1	100%	100%	99.2%	90.4%	71.4%	Intellectual developmental disorder, X-linked 21, 300143

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IL1RN	100%	100%	100%	100%	99.6%	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852
IL2	100%	100%	100%	100%	100%	
IL21	100%	100%	100%	100%	99.8%	?Immunodeficiency, common variable, 11, 615767
IL21R	100%	100%	100%	100%	99.2%	Immunodeficiency 56, 615207
IL27RA	100%	100%	100%	99.9%	99.2%	Immunodeficiency 134 (Epstein-Barr virus-specific), 621405
IL2RA	100%	100%	100%	100%	99.7%	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367;{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942
IL2RB	96.1%	96.1%	100%	100%	99.7%	Immunodeficiency 63 with lymphoproliferation and autoimmunity, 618495
IL2RG	100%	100%	98.8%	90.9%	72.5%	Combined immunodeficiency, X-linked, moderate, 312863;Severe combined immunodeficiency, X-linked, 300400
IL31RA	100%	100%	100%	100%	99.8%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100%	100%	100%	100%	99.9%	Psoriasis 14, pustular, 614204

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IL37	100%	100%	100%	100%	99.7%	?Inflammatory bowel disease (infantile ulcerative colitis) 31, 619398
IL6R	86.3%	86.3%	100%	99.9%	99.4%	[Interleukin 6, serum level of, QTL], 614752;Hyper-IgE syndrome 5, autosomal recessive, with recurrent infections, 618944;[Interleukin-6 receptor, soluble, serum level of, QTL], 614689
IL6ST	96.7%	96.7%	100%	100%	99.6%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
IL7	100%	100%	100%	100%	99.8%	Immunodeficiency 130 with HPV-related verrucosis, 618309
IL7R	100%	100%	100%	100%	99.7%	Immunodeficiency 104, severe combined, 608971
ILDR1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 42, 609646
ILK	100%	100%	100%	100%	99.5%	
IMPA1	81.6%	81.6%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 59, 617323
IMPDH1	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 10, 180105;Leber congenital amaurosis 11, 613837

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IMPG1	100%	100%	100%	100%	99.7%	Macular dystrophy, vitelliform, 4, 616151;Retinitis pigmentosa 91, 153870
IMPG2	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 56, 613581;Macular dystrophy, vitelliform, 5, 616152
INCENP	100%	100%	100%	100%	99.7%	
INF2	100%	100%	99.9%	99.5%	98.1%	Glomerulosclerosis, focal segmental, 5, 613237;Charcot-Marie-Tooth disease, dominant intermediate E, 614455
ING1	100%	100%	100%	100%	99%	Squamous cell carcinoma, head and neck, somatic, 275355
INO80	100%	100%	100%	100%	99.7%	
INPP4A	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech, 621354
INPP5E	100%	100%	100%	99.8%	98.1%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INPP5K	100%	98.5%	100%	100%	99.8%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INPPL1	100%	100%	100%	99.9%	99.4%	Opsismodysplasia, 258480

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

INS	100%	100%	100%	100%	99.1%	Diabetes mellitus, insulin-dependent, 2, 125852;Maturity-onset diabetes of the young, type 10, 613370;Hyperproinsulinemia, 616214;Diabetes mellitus, permanent neonatal 4, 618858
INSC	94.4%	94.4%	100%	100%	99.5%	
INSL3	78.8%	78.8%	100%	100%	99.1%	Cryptorchidism, 219050
INSR	100%	100%	100%	100%	99.5%	Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
INTS1	99.6%	98.7%	100%	100%	99.4%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS11	100%	100%	100%	99.9%	98.6%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
INTS13	100%	100%	100%	100%	99.9%	
INTS6	100%	100%	100%	100%	99.7%	
INTS8	100%	100%	100%	100%	99.4%	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572
INTU	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

INVS	100%	100%	100%	100%	99.9%	Nephronophthisis 2, infantile, 602088
IPMK	100%	100%	100%	99.9%	99.4%	
IPO8	97%	97%	100%	100%	99.8%	VISS syndrome, 619472
IQCB1	95.8%	95.8%	100%	100%	99.7%	Senior-Loken syndrome 5, 609254
IQCE	100%	100%	100%	99.9%	99%	Polydactyly, postaxial, type A7, 617642
IQCN	100%	100%	100%	100%	99.4%	Spermatogenic failure 78, 620170
IQGAP1	100%	100%	100%	100%	99.4%	
IQSEC1	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99.1%	97.9%	97.5%	86%	67.2%	Intellectual developmental disorder, X-linked 1, 309530;Neurodevelopmental disorder, X-linked, with poor or absent speech and behavioral abnormalities, 301164
IQUB	100%	100%	100%	100%	99.6%	
IRAK1	100%	100%	98.2%	89.2%	68.5%	
IRAK4	100%	100%	100%	100%	99.9%	Immunodeficiency 67, 607676
IREB2	98.6%	98.6%	100%	100%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IRF1	100%	100%	100%	100%	99.6%	Nonsmall cell lung cancer, somatic, 211980;Gastric cancer, somatic, 613659;Immunodeficiency 117, mycobacteriosis, autosomal recessive, 620668
IRF2BP2	100%	100%	100%	99.6%	98%	?Immunodeficiency, common variable, 14, 617765
IRF2BPL	100%	100%	99.8%	98.6%	94.6%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRF3	100%	100%	100%	100%	99.4%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 7}, 616532
IRF4	100%	100%	100%	99.9%	99.1%	[Skin/hair/eye pigmentation, variation in, 8], 611724;Immunodeficiency 131, 621097
IRF6	100%	100%	100%	100%	99.4%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300
IRF7	100%	100%	100%	100%	99.2%	Immunodeficiency 39, 616345
IRF8	100%	100%	100%	100%	99.8%	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893;Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 226990
IRF9	100%	100%	100%	99.9%	99%	Immunodeficiency 65, susceptibility to viral infections, 618648

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IRGM	100%	100%	100%	100%	100%	{Mycobacterium tuberculosis, protection against}, 607948;{Inflammatory bowel disease (Crohn disease) 19}, 612278
IRS4	100%	100%	97.5%	85.4%	65.3%	Hypothyroidism, congenital, nongoitrous, 9, 301035
IRX1	100%	100%	100%	99.9%	98%	
IRX5	100%	100%	99.9%	99.6%	97.1%	Hamamy syndrome, 611174
ISCA1	92.4%	92.4%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 5, 617613
ISCA2	100%	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ISCU	100%	100%	100%	100%	99.7%	Myopathy with lactic acidosis, hereditary, 255125
ISG15	100%	100%	100%	100%	99.6%	Immunodeficiency 38, 616126
ISL1	100%	100%	100%	100%	99.8%	
ITCH	92.6%	92.6%	100%	100%	99.8%	Autoimmune disease, multisystem, with facial dysmorphism, 613385
ITGA2	100%	100%	100%	100%	99.9%	Fetomaternal alloimmune thrombocytopenia 3, 621267
ITGA2B	100%	100%	100%	100%	99.4%	Fetomaternal alloimmune thrombocytopenia 2, 621266;Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ITGA3	100%	100%	100%	100%	99.5%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	100%	100%	100%	100%	99.8%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817
ITGA7	100%	99.9%	100%	100%	99.5%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204
ITGA8	100%	100%	100%	100%	99.7%	Renal hypodysplasia/aplasia 1, 191830
ITGAV	99.5%	99.5%	100%	100%	99.8%	Immune dysregulation, neurodevelopmental defects, and colitis, 621375
ITGB2	100%	100%	100%	99.9%	99.7%	Leukocyte adhesion deficiency, 116920
ITGB3	100%	100%	100%	100%	99.5%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;Glanzmann thrombasthenia 2, 619267;Fetomaternal alloimmune thrombocytopenia 1, 621264
ITGB4	100%	99.3%	100%	100%	99.4%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	96.2%	96.2%	100%	100%	99.9%	Amelogenesis imperfecta, type IH, 616221
ITK	100%	100%	100%	100%	99.6%	Lymphoproliferative syndrome 1, 613011

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ITM2B	91.5%	91.5%	100%	100%	99.8%	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079;Dementia, familial British, 176500;Dementia, familial Danish, 117300
ITPA	76.9%	76.9%	100%	99.9%	99.4%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
ITPKB	100%	100%	100%	100%	99.5%	
ITPR1	100%	100%	100%	100%	99.6%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
ITPR2	100%	100%	100%	100%	99.8%	?Anhidrosis, isolated, with normal sweat glands, 106190
ITPR3	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, demyelinating, type 1J, 620111;Immunodeficiency 133 with ectodermal dysplasia with or without peripheral neuropathy, 621254
ITPRID2	100%	100%	100%	100%	99.7%	
ITSN1	100%	100%	100%	100%	99.7%	
ITSN2	100%	100%	100%	100%	99.7%	
IVD	90%	90%	100%	100%	99.6%	Isovaleric acidemia, 243500
IVNS1ABP	100%	100%	100%	100%	99.8%	Immunodeficiency 70, 618969
IYD	89.2%	89.2%	100%	100%	99.8%	Thyroid dyshormonogenesis 4, 274800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

JAG1	100%	100%	100%	100%	99.6%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
JAG2	100%	100%	100%	100%	99.1%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAGN1	100%	100%	100%	100%	99.6%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK1	100%	100%	100%	100%	99.4%	Autoinflammation, immune dysregulation, and eosinophilia, 618999
JAK2	100%	100%	100%	99.9%	99.2%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300
JAK3	100%	100%	100%	99.9%	98.9%	Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type, 600802
JAM2	87.3%	87.3%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	98.1%	93.6%	100%	100%	99.5%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

JARID2	100%	100%	100%	100%	99.5%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098
JKAMP	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures and impaired intellectual and language development, 621533
JMJD1C	100%	100%	100%	100%	99.6%	
JPH1	100%	100%	100%	99.9%	99.2%	Congenital myopathy 25, 620964;{?Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, modifier of}, 607831
JPH2	92.6%	92.6%	100%	99.9%	98.5%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
JPH3	100%	100%	100%	99.9%	98.1%	Huntington disease-like 2, 606438
JUP	100%	100%	100%	100%	99.5%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KALRN	98.3%	98.3%	100%	100%	99.4%	
KANK1	98.1%	98.1%	100%	100%	99.7%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANK2	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KANSL1	98.1%	98.1%	100%	100%	99.8%	Koolen-De Vries syndrome, 610443

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KARS1	94.3%	94.3%	100%	100%	99.8%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KASH5	100%	100%	100%	100%	99.5%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KAT5	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100%	100%	100%	100%	99.7%	Arboleda-Tham syndrome, 616268
KAT6B	100%	100%	100%	100%	99.5%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KAT8	100%	100%	100%	100%	99.5%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100%	100%	100%	100%	99.6%	Lissencephaly 6, with microcephaly, 616212
KATNIP	100%	100%	100%	100%	99.4%	Joubert syndrome 26, 616784
KBTBD13	100%	100%	100%	100%	99.1%	Nemaline myopathy 6, autosomal dominant, 609273
KBTBD2	100%	100%	100%	100%	99.9%	
KCNA1	100%	100%	100%	100%	99.5%	Episodic ataxia/myokymia syndrome, 160120

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KCNA2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100%	100%	100%	99.9%	99.2%	
KCNA4	100%	100%	100%	100%	99.8%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNA5	100%	100%	100%	99.9%	99.3%	Atrial fibrillation, familial, 7, 612240
KCNB1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 26, 616056
KCNB2	100%	100%	100%	100%	99.5%	
KCNC1	100%	100%	100%	99.8%	99%	Epilepsy, progressive myoclonic 7, 616187
KCNC2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 103, 619913
KCNC3	100%	99.5%	99.8%	98.4%	93.4%	Spinocerebellar ataxia 13, 605259
KCND1	100%	100%	98.6%	86.3%	67.7%	
KCND2	100%	100%	100%	99.9%	99.7%	
KCND3	100%	100%	100%	99.9%	99%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNE1	100%	100%	100%	100%	100%	Jervell and Lange-Nielsen syndrome 2, 612347;Long QT syndrome 5, 613695
KCNE2	100%	100%	100%	100%	99.7%	Long QT syndrome 6, 613693;Atrial fibrillation, familial, 4, 611493
KCNE3	100%	100%	100%	100%	99.8%	?Brugada syndrome 6, 613119

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KCNE4	100%	100%	100%	100%	99.4%	
KCNE5	100%	100%	99.1%	90.7%	71.9%	
KCNH1	98.6%	98.6%	100%	100%	99.8%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNH2	100%	100%	100%	100%	99%	Short QT syndrome 1, 609620;Long QT syndrome 2, 613688
KCNH5	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 112, 620537
KCNJ1	100%	100%	100%	100%	99.9%	Bartter syndrome, type 2, 241200
KCNJ10	100%	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNJ11	100%	100%	100%	100%	99.5%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ13	100%	100%	100%	100%	99.5%	Snowflake vitreoretinal degeneration, 193230;Leber congenital amaurosis 16, 614186
KCNJ16	100%	100%	100%	100%	99.9%	Hypokalemic tubulopathy and deafness, 619406

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KCNJ2	100%	100%	100%	100%	99.8%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNJ5	100%	100%	100%	100%	99.2%	Long QT syndrome 13, 613485;Hyperaldosteronism, familial, type III, 613677
KCNJ6	100%	100%	100%	100%	99.8%	Keppen-Lubinsky syndrome, 614098
KCNJ8	89.3%	89.3%	100%	100%	99.7%	
KCNK3	100%	100%	100%	100%	99%	Developmental delay with sleep apnea, 621402;Pulmonary hypertension, primary, 4, 615344
KCNK4	99.4%	99.4%	100%	100%	99.3%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	100%	100%	100%	100%	99.8%	Birk-Barel syndrome, 612292
KCNMA1	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KCNN2	100%	100%	100%	99.9%	98.9%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCNN3	100%	100%	100%	99.9%	99%	Zimmermann-Laband syndrome 3, 618658
KCNN4	96.2%	96.2%	100%	100%	99.3%	Dehydrated hereditary stomatocytosis 2, 616689
KCNQ1	100%	100%	100%	99.9%	99.3%	Short QT syndrome 2, 609621;Atrial fibrillation, familial, 3, 607554;Long QT syndrome 1, 192500;{Long QT syndrome 1, acquired, susceptibility to}, 192500;Jervell and Lange-Nielsen syndrome, 220400
KCNQ1OT1						Beckwith-Wiedemann syndrome, 130650
KCNQ2	94.3%	94.1%	100%	100%	99.3%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200
KCNQ3	100%	100%	100%	100%	99.6%	Seizures, benign neonatal, 2, 121201
KCNQ4	95.3%	93%	100%	99.9%	98.9%	Deafness, autosomal dominant 2A, 600101
KCNQ5	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 46, 617601

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KCNT1	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 57, 617771
KCNU1	100%	100%	100%	100%	99.8%	Spermatogenic failure 79, 620196
KCNV2	100%	100%	100%	100%	99.7%	Cone dystrophy with supernormal rod responses, 610356
KCTD1	100%	100%	100%	99.8%	98.3%	Scalp-ear-nipple syndrome, 181270;Dental radicular dysplasia, 621434
KCTD17	100%	100%	100%	100%	99.1%	Dystonia 26, myoclonic, 616398
KCTD19	100%	100%	100%	100%	99.6%	
KCTD3	100%	100%	100%	100%	99.8%	
KCTD7	100%	100%	100%	100%	99.9%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDELR2	86.8%	86.8%	100%	100%	99.7%	Osteogenesis imperfecta, type XXI, 619131
KDF1	100%	100%	100%	100%	98.8%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/ nail type, 617337
KDM1A	93.5%	93.5%	100%	100%	99.6%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KDM2A	100%	100%	100%	100%	99.5%	
KDM2B	95.9%	94%	100%	99.9%	98.8%	Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities, 621474
KDM3B	100%	100%	100%	100%	99.6%	Diets-Jongmans syndrome, 618846
KDM4B	100%	100%	100%	99.9%	99.2%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5A	100%	100%	100%	100%	99.8%	El Hayek-Chahrour neurodevelopmental syndrome, 620820
KDM5B	97.6%	96.9%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	97.9%	97.8%	98.9%	88.7%	69.9%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM5D	48.9%	48.9%	48.9%	39.3%	20.4%	
KDM6A	100%	100%	99%	91.6%	72.4%	Kabuki syndrome 2, 300867
KDM6B	100%	100%	100%	100%	98.9%	Stolerman neurodevelopmental syndrome, 618505
KDR	100%	100%	100%	100%	99.7%	{Hemangioma, capillary infantile, susceptibility to}, 602089;Hemangioma, capillary infantile, somatic, 602089
KDSR	91.6%	91.6%	100%	100%	98.4%	Erythrokeratodermia variabilis et progressiva 4, 617526

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KERA	100%	100%	100%	99.7%	98.6%	Cornea plana 2, autosomal recessive, 217300
KGD4	100%	100%	100%	99.8%	99.5%	
KHDC3L	100%	100%	100%	100%	99.8%	Hydatidiform mole, recurrent, 2, 614293
KHK	100%	100%	100%	100%	99.7%	?[Fructosuria, essential], 229800
KIAA0586	95.6%	95.6%	100%	100%	99.7%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	100%	99.8%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIAA0825	100%	100%	100%	99.9%	99.6%	Polydactyly, postaxial, type A10, 618498
KIAA1549	99.5%	99.3%	100%	100%	99.3%	Retinitis pigmentosa 86, 618613
KICS2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 83, 621100
KIDINS220	100%	100%	100%	100%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF11	100%	100%	100%	100%	99.8%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KIF12	100%	100%	100%	100%	99.3%	Cholestasis, progressive familial intrahepatic, 8, 619662
KIF14	97.6%	97.6%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF15	99.2%	99.2%	100%	100%	99.7%	?Braddock-Carey syndrome 2, 619981
KIF18A	100%	100%	100%	100%	99.9%	
KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF1B	94.9%	94.9%	100%	100%	99.6%	{Neuroblastoma, susceptibility to, 1}, 256700;Charcot-Marie-Tooth disease, type 2A1, 118210
KIF1C	100%	100%	100%	100%	99.5%	Spastic ataxia 2, autosomal recessive, 611302
KIF20A	100%	100%	100%	100%	99.7%	?Cardiomyopathy, familial restrictive, 6, 619433
KIF21A	100%	100%	100%	100%	99.6%	Fibrosis of extraocular muscles, congenital, 3B, 135700;Fibrosis of extraocular muscles, congenital, 1, 135700
KIF21B	100%	100%	100%	100%	99.5%	
KIF22	99.6%	97.8%	100%	100%	99.7%	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KIF23	97.7%	97.7%	100%	100%	99.7%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KIF24	100%	100%	100%	100%	99.6%	
KIF26A	97.6%	97.6%	100%	99.9%	99.4%	Cortical dysplasia, complex, with other brain malformations 11, 620156
KIF2A	97.9%	97.1%	100%	99.9%	99.6%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 89, 618955
KIF4A	96.1%	96.1%	99.1%	91.6%	73.6%	Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100%	100%	100%	100%	99.5%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
KIF5B	98.8%	96.7%	100%	100%	99.6%	
KIF5C	100%	100%	100%	100%	99.5%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100%	100%	100%	100%	99.2%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KIFBP	95.9%	95.9%	100%	99.9%	99.5%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL1	100%	100%	100%	100%	99.2%	Nephrotic syndrome, type 23, 619201
KIRREL3	100%	100%	100%	100%	99.2%	
KISS1	100%	100%	100%	100%	99.5%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	100%	100%	100%	98.8%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KIT	100%	100%	100%	100%	99.8%	Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626
KITLG	100%	100%	100%	100%	99.9%	Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KIZ	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 69, 615780
KL	100%	99.8%	99.8%	99.3%	98.4%	?Tumoral calcinosis, hyperphosphatemic, familial, 3, 617994

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KLB	100%	100%	100%	100%	99.8%	
KLC2	100%	100%	100%	100%	99.4%	Spastic paraplegia, optic atrophy, and neuropathy, 609541
KLC4	100%	100%	100%	100%	99.5%	?Neurodegeneration, early-childhood-onset, with retinitis pigmentosa, sensorineural hearing loss, and demyelinating peripheral neuropathy, 621129
KLF1	100%	100%	100%	99.9%	98.6%	Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969
KLF11	100%	100%	100%	99.9%	99.6%	Maturity-onset diabetes of the young, type VII, 610508
KLF13	100%	100%	99.9%	99.2%	96.1%	
KLF4	100%	100%	100%	100%	99.3%	
KLF6	100%	100%	100%	100%	99.7%	Gastric cancer, somatic, 613659;Prostate cancer, somatic, 176807
KLF7	100%	100%	100%	100%	99%	
KLHL10	100%	100%	100%	100%	99.8%	Spermatogenic failure 11, 615081
KLHL13	100%	100%	99.3%	93%	75.6%	
KLHL15	89.1%	89.1%	99.3%	93.2%	74.9%	Intellectual developmental disorder, X-linked 103, 300982

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KLHL20	93.7%	93.7%	100%	100%	99.9%	Neurodevelopmental disorder with early-onset seizures, facial dysmorphism, and behavioral abnormalities, 621390
KLHL24	91.5%	91.3%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KLHL3	100%	100%	100%	100%	99.6%	Pseudohypoaldosteronism, type IID, 614495
KLHL40	100%	100%	100%	99.9%	99.5%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	100%	100%	99.7%	Nemaline myopathy 9, 615731
KLHL7	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055
KLHL9	100%	100%	100%	100%	99.7%	
KLK11	100%	100%	100%	100%	98.8%	Ichthyosis with erythrokeratoderma, 620507
KLK4	100%	100%	100%	99.9%	99.2%	Amelogenesis imperfecta, type IIA1, 204700
KLKB1	100%	100%	100%	100%	99.8%	Fletcher factor (prekallikrein) deficiency, 612423
KLLN	100%	100%	100%	100%	99.2%	Cowden syndrome 4, 615107
KMT2A	99.2%	99.2%	100%	100%	99.5%	Wiedemann-Steiner syndrome, 605130

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KMT2B	100%	99.7%	100%	99.9%	98.8%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KMT2C	100%	100%	100%	100%	99.7%	Kleefstra syndrome 2, 617768
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KMT2E	100%	100%	100%	100%	99.5%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNG1	100%	100%	100%	100%	99.7%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960
KNL1	98.7%	98.7%	100%	100%	99.8%	Microcephaly 4, primary, autosomal recessive, 604321
KNSTRN	100%	100%	100%	100%	99.6%	?Roifman-Chitayat syndrome, digenic, 613328
KPNA3	100%	100%	100%	100%	99.7%	Spastic paraplegia 88, autosomal dominant, 620106
KPNA7	100%	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 17, 620319
KPTN	100%	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 41, 615637

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KRAS	100%	100%	100%	100%	100%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800
KREMEN1	85.3%	83.4%	100%	100%	99.2%	Ectodermal dysplasia 13, hair/tooth type, 617392
KRIT1	100%	100%	100%	100%	99.9%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KRT1	100%	100%	100%	100%	99%	Ichthyosis, annular epidermolytic 2, 620148;Palmoplantar keratoderma, nonepidermolytic, 600962;Epidermolytic hyperkeratosis 1, 113800;Palmoplantar keratoderma, epidermolytic, 2, 620411;Keratosis palmoplantaris striata III, 607654;Ichthyosis histrix, Curth-Macklin type, 146590
KRT10	100%	100%	100%	100%	99.3%	Ichthyosis, annular epidermolytic 1, 607602;Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707;Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150;?Ichthyosis histrix, Lambert type, 146600;Ichthyosis with confetti, 609165
KRT12	100%	100%	100%	100%	99.7%	Meesmann corneal dystrophy 1, 122100
KRT13	100%	100%	100%	100%	99.2%	White sponge nevus 2, 615785
KRT14	100%	100%	100%	100%	99.3%	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001;Epidermolysis bullosa simplex 1C, localized, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex 1A, generalized severe, 131760;Naegeli-France schetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KRT16	100%	100%	100%	100%	99.3%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200
KRT17	100%	100%	100%	99.9%	98.9%	Steatocystoma multiplex, 184500;Pachyonychia congenita 2, 167210
KRT18	100%	100%	100%	100%	99.4%	Cirrhosis, cryptogenic, 215600;{Cirrhosis, noncryptogenic, susceptibility to}, 215600
KRT2	100%	100%	100%	100%	99.7%	Ichthyosis bullosa of Siemens, 146800
KRT25	100%	100%	100%	100%	99.7%	Woolly hair, autosomal recessive 3, 616760
KRT3	100%	100%	100%	100%	99.5%	Meesmann corneal dystrophy 2, 618767
KRT31	100%	100%	100%	99.9%	99.4%	
KRT4	100%	100%	100%	99.9%	98.8%	White sponge nevus 1, 193900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KRT5	100%	100%	100%	100%	99.1%	Epidermolysis bullosa simplex 2A, generalized severe, 619555;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599;Epidermolysis bullosa simplex 2B, generalized intermediate, 619588;Epidermolysis bullosa simplex 2C, localized, 619594;Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352
KRT6A	100%	100%	100%	100%	99.7%	Pachyonychia congenita 3, 615726
KRT6B	100%	100%	100%	100%	99.7%	Pachyonychia congenita 4, 615728
KRT6C	99.9%	99.7%	98.7%	97%	93%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100%	100%	100%	100%	99.8%	?Hypotrichosis 13, 615896
KRT74	100%	100%	100%	100%	99.7%	Woolly hair, autosomal dominant, 194300;?Hypotrichosis 3, 613981;?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	100%	100%	100%	100%	99.6%	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT8	100%	100%	100%	100%	99.3%	
KRT81	100%	100%	100%	100%	98.6%	Monilethrix 2, 621169

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KRT82	100%	100%	100%	100%	99.5%	
KRT83	100%	100%	100%	100%	99.3%	Monilethrix 3, 621170;Erythrokeratodermia variabilis et progressiva 5, 617756
KRT85	100%	100%	100%	100%	99.6%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	100%	100%	100%	100%	99.8%	Monilethrix 1, 158000
KRT9	100%	100%	100%	99.9%	99.4%	Palmoplantar keratoderma, epidermolytic, 1, 144200
KY	100%	100%	100%	100%	99.1%	Myopathy, myofibrillar, 7, 617114
KYNU	87.7%	86.6%	100%	100%	99.9%	?Hydroxykynureninuria, 236800;Vertebral, cardiac, renal, and limb defects syndrome 2, 617661
L1CAM	100%	100%	98.4%	88.2%	69.2%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
L2HGDH	92.8%	92.8%	100%	100%	99.6%	L-2-hydroxyglutaric aciduria, 236792
LACC1	94.1%	94.1%	100%	100%	99.7%	Juvenile arthritis, 618795
LACTB	100%	100%	100%	100%	99.6%	
LAGE3	100%	100%	98.7%	89%	67.1%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	98.9%	98.9%	100%	100%	99.6%	Poretti-Boltshauser syndrome, 615960

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LAMA2	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMA3	100%	100%	100%	100%	99.6%	Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784
LAMA4	100%	100%	100%	100%	99.9%	Cardiomyopathy, dilated, 1JJ, 615235
LAMA5	100%	100%	100%	99.9%	99.2%	Nephrotic syndrome, type 26, 620049;?Bent bone dysplasia syndrome 2, 620076
LAMB1	100%	100%	100%	100%	99.7%	Leukoencephalopathy without lacunae, adult-onset, 621424;Leukoencephalopathy with variable cortical brain malformations and/or hydrocephalus, 615191
LAMB2	100%	100%	100%	100%	99.7%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LAMB3	100%	100%	100%	99.9%	99.6%	Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LAMC2	100%	100%	100%	99.9%	99.5%	Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMC3	100%	100%	100%	100%	99.4%	Cortical malformations, occipital, 614115
LAMP2	70.7%	70.7%	98.9%	91%	71.6%	Danon disease, 300257
LAMTOR2	96.6%	85.3%	100%	100%	99.3%	?Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LAPTM5	100%	100%	100%	99.9%	99.5%	
LARGE1	96.9%	96.9%	100%	100%	99.6%	Muscular dystrophy-dystroglycan opathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycan opathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP1	100%	100%	100%	100%	99.2%	
LARP7	100%	100%	100%	100%	99.8%	Alazami syndrome, 615071
LARS1	94.3%	94.3%	100%	100%	99.7%	?Infantile liver failure syndrome 1, 615438
LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	96%	96%	98.3%	88.6%	69.8%	Wilson-Turner syndrome, 309585
LAT	100%	100%	100%	100%	99.7%	Immunodeficiency 52, 617514

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LBR	100%	100%	100%	100%	99.7%	Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140
LBX1	100%	100%	100%	100%	99.1%	?Central hypoventilation syndrome, congenital, 3, 619483
LCA5	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 5, 604537
LCAT	100%	100%	100%	99.9%	99%	Fish-eye disease, 136120;Norum disease, 245900
LCK	98%	98%	100%	100%	99.7%	Immunodeficiency 22, 615758
LCP1	100%	100%	100%	100%	100%	
LCP2	100%	100%	100%	100%	99.6%	Immunodeficiency 81, 619374
LCT	100%	100%	100%	100%	99.5%	Lactase deficiency, congenital, 223000
LDB1	100%	100%	100%	99.9%	99.5%	
LDB3	95.9%	94.9%	100%	100%	99.4%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, dilated, 2L, 621237;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493
LDHA	100%	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933
LDHB	100%	100%	100%	100%	99.5%	[Lactate dehydrogenase-B deficiency], 614128

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LDHD	100%	100%	100%	100%	99.5%	D-lactic aciduria with susceptibility to gout, 245450
LDLR	100%	100%	100%	100%	99.1%	LDL cholesterol level QTL2, 143890;Hypercholesterolemia, familial, 1, 143890
LDLRAP1	93.2%	90.2%	100%	100%	99.2%	Hypercholesterolemia, familial, 4, 603813
LEF1	99.5%	97.2%	100%	100%	99.6%	Ectodermal dysplasia 17 with or without limb malformations, 621224
LEMD2	100%	100%	100%	100%	99.8%	Marbach-Rustad progeroid syndrome, 619322;Cataract 46, juvenile-onset, 212500
LEMD3	100%	100%	100%	99.9%	99%	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
LEP	100%	100%	100%	100%	99.5%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100%	100%	99.7%	Obesity, morbid, due to leptin receptor deficiency, 614963
LETM1	92.4%	89.8%	100%	100%	99.5%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LFNG	99.9%	97.1%	100%	99.9%	98.8%	Spondylocostal dysostosis 3, autosomal recessive, 609813
LGI1	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 121, 621475;Epilepsy, familial temporal lobe, 1, 600512

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LGI3	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007
LGI4	100%	100%	100%	99.9%	99.4%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LGR4	100%	100%	100%	99.9%	99.4%	Delayed puberty, self-limited, 619613;{Bone mineral density, low, susceptibility to}, 615311
LHB	100%	100%	100%	99.8%	98%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100%	100%	100%	100%	99.7%	Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410
LHFPL5	100%	100%	100%	99.8%	99.2%	Deafness, autosomal recessive 67, 610265
LHX1	100%	100%	100%	99.9%	98.5%	
LHX2	100%	100%	100%	99.9%	99.3%	
LHX3	100%	100%	100%	99.6%	98.9%	Pituitary hormone deficiency, combined, 3, 221750
LHX4	100%	100%	100%	100%	99.2%	Pituitary hormone deficiency, combined, 4, 262700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LHX8	92.5%	92.5%	100%	100%	99.1%	
LIAS	100%	100%	100%	100%	99.9%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIFR	100%	100%	100%	100%	99.7%	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559
LIG1	100%	100%	100%	99.9%	99.1%	Immunodeficiency 96, 619774
LIG3	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780
LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LIM2	100%	100%	100%	100%	98.6%	Cataract 19, multiple types, 615277
LIMS2	100%	99.9%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue, 616827
LINGO1	100%	100%	100%	99.9%	98.1%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 27, 614340
LIPA	95.6%	95.2%	100%	100%	99.9%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LIPC	100%	100%	100%	100%	99.6%	{Diabetes mellitus, noninsulin-dependent}, 125853;Hepatic lipase deficiency, 614025;[High density lipoprotein cholesterol level QTL 12], 612797

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LIPE	100%	100%	100%	100%	99.4%	Lipodystrophy, familial partial, type 6, 615980
LIPH	100%	100%	100%	100%	99.6%	Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 8, 613943
LIPT1	100%	100%	100%	100%	99.8%	Lipoyltransferase 1 deficiency, 616299
LIPT2	100%	100%	100%	100%	99%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LITAF	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, type 1C, 601098
LMAN1	100%	100%	100%	100%	99.7%	Combined factor V and VIII deficiency, 227300
LMAN2L	100%	100%	100%	100%	99.9%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887
LMBR1	96.1%	95.5%	100%	100%	99.7%	Syndactyly, type IV, 186200;Laurin-Sandrow syndrome, 135750;Acheiropody, 200500;Triphalangeal thumb-polysyndactyly syndrome, 190605
LMBRD1	86.4%	86.4%	100%	99.9%	99.6%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMBRD2	100%	100%	100%	100%	99.9%	Developmental delay with variable neurologic and brain abnormalities, 619694

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LMCD1	100%	100%	100%	99.9%	99.3%	
LMF1	91.5%	91.5%	100%	100%	99.8%	Lipase deficiency, combined, 246650
LMNA	100%	100%	100%	99.9%	99.3%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMNB1	100%	100%	100%	99.9%	99.2%	Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	92.9%	92.9%	100%	99.8%	98.3%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709
LMOD1	100%	100%	100%	100%	98.8%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LMOD2	100%	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 2G, 619897

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LMOD3	100%	100%	100%	99.9%	99.2%	Nemaline myopathy 10, 616165
LMX1A	100%	100%	100%	99.9%	99.6%	Deafness, autosomal dominant 7, 601412
LMX1B	100%	100%	100%	100%	99%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LNPK	85.8%	85.8%	100%	100%	99.8%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	99.3%	97%	100%	100%	99.5%	CODAS syndrome, 600373
LORICRIN	100%	100%	100%	99.9%	97.4%	Vohwinkel syndrome with ichthyosis, 604117
LOX	100%	100%	100%	99.9%	98.5%	Aortic aneurysm, familial thoracic 10, 617168
LOXHD1	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 77, 613079
LOXL1	100%	100%	100%	99.8%	98.5%	{Exfoliation syndrome, susceptibility to}, 177650
LOXL3	100%	100%	100%	100%	99.3%	Myopia 28, autosomal recessive, 619781
LPAR6	100%	100%	100%	100%	99.9%	Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN1	94%	94%	100%	100%	99.7%	Myoglobinuria, acute recurrent, autosomal recessive, 268200
LPIN2	99.3%	99.2%	100%	99.9%	99.4%	Majeed syndrome, 609628

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LPL	100%	100%	100%	100%	99.6%	Lipoprotein lipase deficiency, 238600;[High density lipoprotein cholesterol level QTL 11], 238600;Combined hyperlipidemia, familial, 144250
LPP	100%	100%	100%	100%	99.2%	Lipoma;Leukemia, acute myeloid, 601626
LRAT	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 14, 613341;Retinal dystrophy, early-onset severe, 613341;Retinitis pigmentosa, juvenile, 613341
LRBA	99.8%	99.8%	100%	100%	99.8%	Immunodeficiency, common variable, 8, with autoimmunity, 614700
LRIF1	100%	100%	100%	100%	99.7%	?Facioscapulohumeral muscular dystrophy 3, digenic, 619477
LRIG2	98%	98%	100%	100%	99.8%	Urofacial syndrome 2, 615112
LRIT3	100%	100%	100%	100%	99.9%	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058
LRMDA	97.8%	97.8%	100%	100%	99.4%	Albinism, oculocutaneous, type VII, 615179
LRP1	100%	100%	100%	100%	99.3%	?Keratosis pilaris atrophicans, 604093;Developmental dysplasia of the hip 3, 620690
LRP12	100%	100%	100%	100%	99.6%	Oculopharyngodistal myopathy 1, 164310;Amyotrophic lateral sclerosis 28, 620452

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRP4	100%	100%	100%	99.9%	99.4%	?Myasthenic syndrome, congenital, 17, 616304;Sclerosteosis 2, 614305;Cenani-Lenz syndactyly syndrome, 212780
LRP5	100%	100%	100%	100%	99%	Osteopetrosis, autosomal dominant 1, 607634;Polycystic liver disease 4 with or without kidney cysts, 617875;Endosteal hyperostosis, 144750;Osteoporosis-pseudoglioma syndrome, 259770;[Bone mineral density variability 1, high bone mass], 601884;Exudative vitreoretinopathy 4, 601813
LRP6	98%	98%	100%	100%	99.6%	Osteopetrosis, autosomal dominant 4, 621449;{Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724;Exudative vitreoretinopathy 8, 621268
LRPAP1	84.8%	84.8%	100%	100%	99.4%	Myopia 23, autosomal recessive, 615431
LRPPRC	96.8%	96.7%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC10	100%	100%	100%	100%	99.8%	
LRRC23	100%	100%	100%	99.9%	99%	Spermatogenic failure 92, 620848

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LRRC32	100%	100%	100%	100%	99.3%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
LRRC56	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 39, 618254
LRRC7	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 77, 621415
LRRC8A	96.1%	96.1%	100%	100%	99.2%	?Agammaglobulinemia 5, 613506
LRRC8C	100%	100%	100%	100%	99.9%	TIMES syndrome, 621056
LRRK1	100%	100%	100%	100%	99.5%	Osteosclerotic metaphyseal dysplasia, 615198
LRRK2	100%	100%	100%	100%	99.7%	{Parkinson disease 8}, 607060
LRSAM1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, axonal, type 2P, 614436
LRTOMT	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 63, 611451
LSM1	100%	100%	100%	100%	99.2%	FICUS syndrome, 621193
LSM11	100%	100%	100%	100%	99.1%	?Aicardi-Goutieres syndrome 8, 619486
LSM7	100%	100%	100%	100%	98.7%	Leukodystrophy and cerebellar atrophy, 621191
LSR	83.5%	83.5%	100%	99.9%	99%	
LSS	100%	100%	100%	100%	99.8%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840
LTBP1	100%	100%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IIE, 619451

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LTBP2	100%	100%	100%	100%	99.7%	Glaucoma 3, primary congenital, D, 613086;Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750;?Weill-Marchesani syndrome 3, recessive, 614819
LTBP3	100%	100%	100%	99.9%	98.9%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LTBP4	100%	100%	100%	100%	99%	Cutis laxa, autosomal recessive, type IC, 613177
LTC4S	100%	100%	99.8%	99.2%	98.3%	Leukotriene C4 synthase deficiency, 614037
LTV1	100%	100%	100%	100%	99.9%	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199
LYN	100%	100%	100%	100%	99.3%	Autoinflammatory disease, systemic, with vasculitis, 620376
LYRM4	68%	68%	100%	100%	99.5%	?Combined oxidative phosphorylation deficiency 19, 615595
LYRM7	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYSET	100%	100%	100%	100%	99.7%	Dysostosis multiplex, Ain-Naz type, 619345
LYST	99.5%	99.5%	100%	100%	99.9%	Chediak-Higashi syndrome, 214500
LYZ	100%	100%	100%	100%	99.8%	Amyloidosis, hereditary systemic 5, 620658
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
LZTS1	100%	100%	100%	99.9%	99.1%	Esophageal squamous cell carcinoma, somatic, 133239
M1AP	100%	100%	100%	99.9%	99.2%	Spermatogenic failure 48, 619108
MAB21L1	100%	100%	100%	100%	99.3%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	100%	100%	99.2%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877
MACF1	100%	100%	100%	100%	99.7%	Lissencephaly 9 with complex brainstem malformation, 618325
MAD1L1	100%	98.9%	100%	100%	99.5%	Prostate cancer, somatic, 176807;Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition, 620189;Lymphoma, B-cell, somatic
MAD2L1BP	86.6%	86.6%	100%	100%	99.5%	
MAD2L2	100%	100%	100%	99.9%	99.1%	?Fanconi anemia, complementation group V, 617243
MADD	98.7%	98.7%	100%	100%	99.5%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004
MAF	95.1%	91.7%	100%	99.4%	96.8%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MAFA	100%	99.9%	100%	99.4%	93.5%	Insulinomatosis and diabetes mellitus, 147630
MAFB	100%	100%	100%	99.9%	98.8%	Duane retraction syndrome 3, 617041;Multicentric carpotarsal osteolysis syndrome, 166300
MAG	100%	100%	100%	100%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MAGED2	99.4%	97.2%	98.6%	89.3%	70.4%	Bartter syndrome, type 5, antenatal, transient, 300971
MAGEL2	100%	100%	100%	100%	99.7%	Schaaf-Yang syndrome, 615547
MAGI2	99.6%	97.9%	100%	99.8%	99%	Nephrotic syndrome, type 15, 617609
MAGT1	93.8%	93.8%	99%	88.7%	70.8%	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031
MAK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 62, 614181
MAL	71.9%	71.9%	100%	100%	99.6%	?Leukodystrophy, hypomyelinating, 28, 620978
MALT1	93.9%	93.9%	100%	100%	99.7%	Immunodeficiency 12, 615468
MAML2	100%	100%	100%	100%	99.7%	Mucoepidermoid salivary gland carcinoma
MAMLD1	100%	100%	98.8%	89.1%	70.8%	Hypospadias 2, X-linked, 300758
MAN1B1	99.8%	98.1%	100%	100%	99.6%	Rafiq syndrome, 614202
MAN2A2	100%	100%	100%	100%	99.4%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MAN2B1	100%	100%	100%	100%	99.1%	Mannosidosis, alpha-, types I and II, 248500
MAN2B2	89.8%	87.3%	100%	100%	99.6%	Congenital disorder of glycosylation type 1EE with or without immunodeficiency, 621140
MAN2C1	98.4%	98.4%	100%	99.9%	99.5%	Congenital disorder of deglycosylation 2, 619775
MANBA	99.4%	97.5%	100%	100%	99.5%	Mannosidosis, beta, 248510
MANF	100%	100%	100%	99.9%	99%	Diabetes, deafness, developmental delay, and short stature syndrome, 620651
MAOA	100%	100%	99.6%	92.1%	73.7%	Brunner syndrome, 300615
MAP1B	100%	100%	100%	100%	99.6%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP1LC3B2	100%	100%	100%	100%	99.4%	
MAP2K1	95.8%	95.8%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	92.4%	92.3%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 4, 615280
MAP2K4	100%	100%	100%	100%	99.7%	
MAP3K1	100%	100%	100%	99.9%	99.5%	46XY sex reversal 6, 613762
MAP3K14	100%	100%	100%	100%	99.5%	Immunodeficiency 112, 620449
MAP3K20	100%	100%	100%	100%	99.9%	Centronuclear myopathy 6 with fiber-type disproportion, 617760;Split-foot malformation with mesoaxial polydactyly, 616890

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MAP3K3	100%	100%	100%	100%	99.7%	Cerebral cavernous malformations 5, 621032
MAP3K7	97.9%	97.9%	100%	100%	99.8%	Frontometaphyseal dysplasia 2, 617137;Cardiospondylocarpofacial syndrome, 157800
MAP3K8	95%	95%	100%	100%	99.5%	Lung cancer, somatic, 211980
MAP4K4	100%	100%	100%	100%	99.7%	
MAPK1	100%	100%	100%	99.9%	99.4%	Noonan syndrome 13, 619087
MAPK8	100%	100%	100%	100%	99.7%	
MAPK8IP3	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK3	92.7%	89.5%	100%	100%	99.7%	?Macular dystrophy, patterned, 3, 617111
MAPKAPK5	100%	100%	100%	100%	99.4%	Neurocardiofaciodigital syndrome, 619869
MAPKBP1	100%	100%	100%	100%	99.8%	Nephronophthisis 20, 617271
MAPRE2	100%	100%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 2, 616734
MAPT	95.4%	95.4%	100%	99.9%	99.2%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MARCHF6	98.7%	98.7%	100%	100%	99.5%	Epilepsy, familial adult myoclonic, 3, 613608

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MARK2	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 76, 621285
MARK3	96.2%	96.2%	100%	100%	99.5%	?Visual impairment and progressive phthisis bulbi, 618283
MARS1	89.3%	88.7%	100%	100%	99.5%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MARS2	100%	100%	100%	99.9%	99.2%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MARVELD2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 49, 610153
MASP1	100%	100%	100%	100%	99.6%	3MC syndrome 1, 257920
MASP2	97.2%	97.2%	100%	100%	99.7%	MASP2 deficiency, 613791
MAST1	100%	100%	100%	100%	99.3%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAST3	100%	100%	100%	99.9%	99%	Developmental and epileptic encephalopathy 108, 620115
MAST4	100%	100%	100%	100%	99.5%	
MASTL	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MAT1A	100%	99.9%	100%	99.9%	99.4%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAT2A	100%	100%	100%	99.9%	99.7%	
MATN3	100%	100%	100%	100%	99.8%	{Osteoarthritis susceptibility 2}, 140600;Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type, 608728;Epiphyseal dysplasia, multiple, 5, 607078
MATR3	100%	100%	100%	100%	99.6%	Amyotrophic lateral sclerosis 21, 606070
MAU2	100%	100%	100%	100%	99.5%	Cornelia de Lange syndrome 7, 621570
MAX	100%	100%	100%	100%	99.7%	Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300
MB	100%	100%	100%	100%	99.4%	Myopathy, sarcoplasmic body, 620286
MBD4	100%	100%	100%	100%	99.9%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MBD5	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBIP	96.6%	96%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MBOAT7	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS1	100%	100%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392;CAOP syndrome, 621252
MBTPS2	94.6%	94.6%	99.3%	91.4%	73.6%	Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918
MC2R	100%	100%	100%	100%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MC4R	100%	100%	100%	100%	99.7%	Obesity (BMIQ20), 618406;{Obesity, resistance to (BMIQ20)}, 618406
MCAT	95.9%	95.9%	100%	100%	99.6%	Optic atrophy 15, 620583
MCC	100%	100%	100%	100%	99.5%	Colorectal cancer, somatic, 114500
MCCC1	94.5%	94.5%	100%	100%	99.8%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	93.5%	93.5%	100%	100%	99.9%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCEE	100%	100%	100%	100%	99.5%	Methylmalonyl-CoA epimerase deficiency, 251120
MCFD2	100%	100%	100%	100%	99.6%	Factor V and factor VIII, combined deficiency of, 613625

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MCIDAS	100%	100%	100%	99.9%	98.9%	Ciliary dyskinesia, primary, 42, 618695
MCM10	100%	100%	100%	100%	99.7%	Immunodeficiency 80 with or without cardiomyopathy, 619313
MCM2	100%	100%	100%	100%	99.4%	?Deafness, autosomal dominant 70, 616968
MCM3AP	100%	100%	100%	100%	99.5%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM4	95.4%	95.4%	100%	100%	99.8%	Immunodeficiency 54, 609981
MCM5	96.9%	96.9%	100%	100%	99.6%	?Meier-Gorlin syndrome 8, 617564
MCM6	100%	100%	100%	100%	99.8%	Lactase persistence/nonpersistence, 223100
MCM8	94.4%	94.4%	100%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	100%	99.8%	Ovarian dysgenesis 4, 616185
MCMDC2	100%	100%	100%	100%	99.9%	
MCOLN1	98.3%	95.7%	100%	100%	99.7%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MCPH1	94.6%	92.8%	100%	100%	99.7%	Microcephaly 1, primary, autosomal recessive, 251200
MCTS1	91.5%	91.5%	99.7%	94.4%	75.5%	Immunodeficiency 118, mycobacteriosis, 301115
MCUR1	100%	100%	100%	99.9%	99.4%	
MDFIC	100%	100%	100%	100%	99.8%	Lymphatic malformation 12, 620014
MDGA2	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MDH1	88.5%	88.5%	100%	100%	99.8%	?Developmental and epileptic encephalopathy 88, 618959
MDH2	100%	100%	100%	100%	98.7%	Developmental and epileptic encephalopathy 51, 617339
MDM1	100%	100%	100%	100%	99.6%	
MDM2	94%	94%	100%	100%	99.3%	{Accelerated tumor formation, susceptibility to}, 614401;?Lessel-Kubisch syndrome, 618681
MDM4	100%	100%	100%	100%	99.7%	?Bone marrow failure syndrome 6, 618849
ME2	94.1%	94.1%	100%	100%	99.9%	
MECOM	100%	100%	100%	100%	99.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MECP2	100%	100%	99%	84.8%	65.9%	Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750
MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MED11	100%	100%	100%	100%	99.7%	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327
MED12	100%	99.9%	98.1%	87.5%	68.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED12L	100%	100%	100%	100%	99.7%	Nizon-Isidor syndrome, 618872
MED13	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	100%	100%	100%	100%	99.6%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED16	100%	100%	99.9%	99.6%	97.3%	Guillouet-Gordon syndrome, 621220
MED17	100%	100%	100%	100%	99.6%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100%	100%	100%	100%	99.4%	Basel-Vanagaite-Smirin-Yosef syndrome, 616449
MED27	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MEF2C	100%	100%	100%	100%	99.7%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEFV	96.1%	96.1%	100%	100%	99.6%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MEGF10	100%	100%	100%	100%	99.6%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEI1	100%	100%	100%	100%	99.6%	Hydatidiform mole, recurrent, 3, 618431
MEI4	100%	100%	100%	100%	99.9%	
MEIOB	100%	100%	100%	100%	99.9%	Premature ovarian failure 23, 620686;Spermatogenic failure 22, 617706
MEIS2	89.6%	88.5%	100%	100%	99.6%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
MEN1	100%	100%	100%	100%	99.1%	Lipoma, somatic;Angiofibroma, somatic;Multiple endocrine neoplasia 1, 131100;Carcinoid tumor of lung;Adrenal adenoma, somatic;Parathyroid adenoma, somatic
MEOX1	100%	100%	100%	100%	98.8%	Klippel-Feil syndrome 2, 214300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MERTK	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 38, 613862
MESD	91.8%	91.8%	100%	99.9%	99.1%	Osteogenesis imperfecta, type XX, 618644
MESP1	100%	100%	100%	99.9%	99.3%	
MESP2	100%	99.9%	100%	100%	99.8%	Spondylocostal dysostosis 2, autosomal recessive, 608681
MET	100%	100%	100%	100%	99.8%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
METTL13	100%	100%	100%	100%	99.7%	{?Deafness, autosomal recessive 26, modifier of}, 605429
METTL23	100%	100%	100%	100%	100%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFAP5	100%	100%	100%	100%	99.7%	Aortic aneurysm, familial thoracic 9, 616166
MFF	89.1%	89.1%	100%	100%	99.9%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MFN2	100%	100%	100%	100%	99.7%	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800;Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260;Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087;Hereditary motor and sensory neuropathy VIA, 601152
MFRP	100%	100%	100%	100%	99.4%	Microphthalmia, isolated 5, 611040;Nanophthalmos 2, 609549
MFSD2A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100%	100%	100%	100%	100%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGA	100%	100%	100%	100%	99.8%	Premature ovarian failure 26, 621065
MGAT2	100%	100%	100%	99.8%	99%	Congenital disorder of glycosylation, type IIa, 212066
MGME1	100%	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 11, 615084
MGP	100%	100%	100%	100%	99.7%	Keutel syndrome, 245150
MGRN1	100%	100%	100%	100%	99.5%	
MIA3	100%	100%	100%	100%	99.7%	?Odontochondrodysplasia 2 with hearing loss and diabetes, 619269

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MIB1	94.8%	94.8%	100%	100%	99.7%	Left ventricular noncompaction 7, 615092
MICOS13	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	86.7%	86.7%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MICU2	88.6%	85.9%	100%	99.9%	99.5%	
MID1	100%	100%	98.8%	88.7%	69.8%	Opitz GBBB syndrome, 300000
MID2	100%	100%	98.9%	90.9%	72.5%	?Intellectual developmental disorder, X-linked 101, 300928
MIDEAS	100%	100%	100%	100%	99.2%	
MIEF1	100%	100%	100%	100%	99.3%	Optic atrophy 14, 620550
MIEF2	100%	100%	100%	100%	99%	?Combined oxidative phosphorylation deficiency 49, 619024
MINAR2	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 120, 620238
MINPP1	100%	100%	100%	100%	99.9%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527
MIP	100%	100%	100%	100%	99.7%	Cataract 15, multiple types, 615274
MIPEP	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 31, 617228
MIR140						Spondyloepiphyseal dysplasia, Nishimura type, 618618
MIR17HG						
MIR184						EDICT syndrome, 614303

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MIR204						Retinal dystrophy and iris coloboma with or without cataract, 616722
MIR96						Deafness, autosomal dominant 50, 613074
MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKRN3	100%	100%	100%	100%	99.7%	Precocious puberty, central, 2, 615346
MKS1	99.1%	99%	100%	100%	99.7%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MLC1	100%	100%	100%	100%	99.2%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLH1	100%	100%	100%	100%	99.7%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLH3	100%	100%	100%	99.9%	99.2%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MLIP	100%	100%	100%	100%	99.7%	Myopathy with myalgia, increased serum creatine kinase, and with or without episodic rhabdomyolysis, 620138
MLLT10	97%	97%	100%	100%	99.7%	Leukemia, acute myeloid, 601626
MLPH	95.4%	95.4%	100%	100%	99.3%	Griscelli syndrome, type 3, 609227
MLYCD	100%	100%	100%	100%	99.4%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	99.8%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	91.8%	91.8%	100%	99.9%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.3%	79.3%	100%	100%	99.7%	Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria -megaloblastic anemia, cblD type, 620952
MME	97.7%	97.7%	100%	100%	99.7%	?Spinocerebellar ataxia 43, 617018;Charcot-Marie-Tooth disease, axonal, type 2T, 617017
MMGT1	100%	100%	99.4%	92.8%	75.4%	
MMP1	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MMP13	92.2%	92.2%	100%	100%	99.4%	?Spondyloepimetaphyseal dysplasia, Missouri type, 602111;Metaphyseal anadysplasia 1, 602111;Metaphyseal dysplasia, Spahr type, 250400
MMP14	94.9%	94.9%	100%	100%	99.5%	Winchester syndrome, 277950
MMP19	100%	100%	100%	100%	99.5%	Cavitary optic disc anomalies, 611543
MMP2	100%	100%	100%	100%	99.5%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA2, 612529
MMP21	100%	100%	100%	100%	99.4%	Heterotaxy, visceral, 7, autosomal, 616749
MMP9	100%	100%	100%	99.9%	98.8%	Metaphyseal anadysplasia 2, 613073
MMS19	96.5%	96.5%	100%	100%	99.7%	
MMUT	100%	100%	100%	100%	99.7%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100%	100%	100%	99.9%	98.5%	CEBALID syndrome, 618774;Meningioma, 607174
MNS1	100%	100%	100%	99.9%	99.8%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
MNX1	98.3%	91.1%	99.8%	98.7%	94.1%	Currarino syndrome, 176450
MOCOS	97.6%	97.6%	100%	100%	99.6%	Xanthinuria, type II, 603592
MOCS1	100%	100%	100%	100%	99.4%	Molybdenum cofactor deficiency A, 252150
MOCS2	100%	100%	100%	100%	99.9%	Molybdenum cofactor deficiency B1, 252160

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MOCS3	100%	100%	100%	100%	99.8%	Molybdenum cofactor deficiency, type B2, 621373
MOG	99.1%	97.5%	100%	100%	99.8%	?Narcolepsy 7, 614250
MOGS	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100%	100%	100%	100%	99.3%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MOS	100%	100%	99.9%	99.8%	98.3%	Oocyte/zygote/embryo maturation arrest 20, 620383
MOV10L1	100%	100%	100%	100%	99.6%	?Spermatogenic failure 73, 619878
MPC1	100%	100%	100%	100%	99.9%	Mitochondrial pyruvate carrier deficiency, 614741
MPC2	100%	100%	100%	99.9%	99.7%	
MPDU1	100%	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100%	100%	100%	100%	99.7%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPEG1	100%	100%	100%	100%	99.5%	Immunodeficiency 77, 619223
MPI	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ib, 602579
MPIG6B	100%	100%	100%	100%	99.9%	?Thrombocytopenia, anemia, and myelofibrosis, 617441

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MPL	100%	100%	100%	99.9%	99.3%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MPLKIP	100%	100%	100%	100%	99%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPO	100%	100%	100%	100%	99.5%	{Alzheimer disease, susceptibility to}, 104300;Myeloperoxidase deficiency, 254600;{Lung cancer, protection against, in smokers}
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MPZ	100%	100%	100%	100%	98.9%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736
MPZL2	87.9%	87.9%	100%	100%	99.4%	Deafness, autosomal recessive 111, 618145
MRAP	100%	100%	100%	100%	99.6%	Glucocorticoid deficiency 2, 607398

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MRAS	100%	100%	100%	100%	99.7%	Noonan syndrome 11, 618499
MRE11	97.4%	97.4%	100%	100%	99.9%	Ataxia-telangiectasia-like disorder 1, 604391
MRM2	87.4%	87.3%	100%	99.7%	98.3%	Mitochondrial DNA depletion syndrome 17, 618567
MRPL12	100%	100%	100%	100%	99.3%	?Combined oxidative phosphorylation deficiency 45, 618951
MRPL24	100%	100%	100%	99.7%	98.7%	
MRPL3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 9, 614582
MRPL34	100%	100%	100%	100%	98.7%	
MRPL39	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 59, 620646
MRPL40	100%	100%	100%	99.9%	99.5%	
MRPL42	71.3%	71.3%	100%	99.9%	99.6%	
MRPL44	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 16, 615395
MRPL49	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 60, 621195
MRPL50	100%	100%	100%	99.6%	98.7%	
MRPL57	100%	100%	100%	100%	100%	
MRPS14	85.2%	85.2%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 38, 618378
MRPS16	100%	100%	100%	100%	99%	Combined oxidative phosphorylation deficiency 2, 610498
MRPS2	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 36, 617950

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MRPS22	86.6%	86.6%	100%	100%	99.9%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS23	100%	100%	100%	100%	99.7%	?Combined oxidative phosphorylation deficiency 46, 618952
MRPS25	74.2%	74.2%	100%	100%	99.8%	?Combined oxidative phosphorylation deficiency 50, 619025
MRPS28	73.8%	73.8%	100%	99.8%	97.8%	?Combined oxidative phosphorylation deficiency 47, 618958
MRPS34	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 32, 617664
MRPS7	100%	100%	100%	99.9%	99%	?Combined oxidative phosphorylation deficiency 34, 617872
MRRF	100%	100%	100%	100%	100%	
MRTFA	100%	100%	100%	100%	99.6%	?Immunodeficiency 66, 618847
MRTFB	100%	100%	100%	100%	99.8%	
MS4A1	100%	100%	100%	100%	99.9%	?Immunodeficiency, common variable, 5, 613495
MSH2	90.6%	90.6%	100%	100%	99.6%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	98%	98%	100%	100%	99.7%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH4	100%	100%	100%	100%	99.8%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MSH5	100%	100%	100%	100%	99.3%	?Premature ovarian failure 13, 617442;Spermatogenic failure 74, 619937
MSH6	100%	100%	100%	100%	99.5%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MSL2	92.7%	92.7%	100%	100%	99.8%	Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985
MSL3	100%	100%	99.3%	91.2%	72.6%	Basilicata-Akhtar syndrome, 301032
MSMO1	100%	100%	100%	100%	99.9%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSN	100%	100%	99.1%	90.7%	73.2%	Immunodeficiency 50, 300988
MSR1	100%	100%	100%	99.4%	98.3%	Barrett esophagus/esophageal adenocarcinoma, 614266
MSRB3	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 74, 613718
MST1R	100%	100%	100%	100%	99.6%	{Nasopharyngeal carcinoma, susceptibility to, 3}, 617075
MSTN	100%	100%	100%	100%	100%	?Muscle hypertrophy, 614160
MSTO1	100%	100%	99.9%	99.1%	97.7%	Myopathy, mitochondrial, and ataxia, 617675
MSX1	100%	100%	100%	99.8%	98.4%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MSX2	100%	100%	100%	99.9%	99%	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500
MTAP	91.5%	91.5%	100%	100%	99.4%	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250
MTERF3	100%	100%	100%	100%	99.9%	
MTFMT	91.8%	91.8%	100%	99.9%	98.6%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFD1	94.9%	92%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780
MTHFR	100%	100%	100%	100%	99.5%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTHFS	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTM1	100%	100%	99.2%	92.2%	74.7%	Myopathy, centronuclear, X-linked, 310400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MTMR2	100%	100%	100%	99.9%	99.6%	Charcot-Marie-Tooth disease, type 4B1, 601382
MTO1	94.8%	91.9%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MTPAP	96.6%	96.6%	100%	100%	99.4%	?Spastic ataxia 4, autosomal recessive, 613672
MTR	100%	100%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100%	100%	100%	100%	99.7%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTSS2	100%	100%	100%	99.9%	99.1%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086
MT-TI	99.6%	95.9%				
MTTP	100%	100%	100%	100%	99.8%	Abetalipoproteinemia, 200100
MTX2	87.1%	87.1%	100%	100%	99.2%	Mandibuloacral dysplasia progeroid syndrome, 619127

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MUC1	100%	100%	100%	99.9%	99.3%	Tubulointerstitial kidney disease, autosomal dominant, 2, 174000
MUC16	100%	100%	100%	100%	99.6%	
MUSK	100%	100%	100%	100%	99.7%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MUTYH	100%	100%	100%	99.9%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
MVD	91.2%	91.2%	100%	100%	99.1%	Porokeratosis 7, multiple types, 614714
MVK	100%	100%	100%	100%	99.8%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MXI1	100%	100%	100%	99.8%	98.6%	Prostate cancer, somatic, 176807;Neurofibrosarcoma, somatic
MYBPC1	100%	100%	100%	100%	99.8%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335
MYBPC3	100%	100%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396
MYBPHL	99.9%	98.7%	100%	100%	99.4%	
MYC	100%	100%	100%	100%	99.4%	Burkitt lymphoma, somatic, 113970
MYCBP2	100%	99.8%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MYCN	100%	100%	100%	100%	98.9%	Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748
MYD88	100%	100%	100%	100%	99.9%	Macroglobulinemia, Waldenstrom, somatic, 153600;Immunodeficiency 68, 612260
MYF5	100%	100%	100%	100%	99.9%	Ophthalmoplegia, external, with rib and vertebral anomalies, 618155
MYH10	99.5%	99.5%	100%	100%	99.6%	
MYH11	100%	100%	100%	100%	99.3%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYH14	100%	100%	100%	99.9%	98.8%	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369;Deafness, autosomal dominant 4A, 600652
MYH2	100%	100%	100%	99.9%	99.8%	Congenital myopathy 6 with ophthalmoplegia, 605637
MYH3	100%	100%	100%	100%	99.5%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MYH6	100%	100%	100%	100%	99.4%	?Atrial septal defect 3, 614089;{Sick sinus syndrome 3}, 614090;Cardiomyopathy, dilated, 1EE, 613252;Cardiomyopathy, hypertrophic, 14, 613251
MYH7	99.1%	99.1%	100%	100%	99.7%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYH7B	100%	100%	100%	100%	99.2%	
MYH8	100%	100%	100%	100%	99.6%	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYH9	97.3%	97%	100%	100%	99.4%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYL1	100%	100%	100%	100%	99.7%	Congenital myopathy 14, 618414
MYL11	100%	100%	100%	100%	99.1%	Arthrogryposis, distal, type 1C, 619110
MYL2	100%	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MYL3	81.6%	79.9%	100%	100%	99.2%	Cardiomyopathy, hypertrophic, 8, 608751
MYL4	100%	100%	100%	99.9%	99.5%	?Atrial fibrillation, familial, 18, 617280
MYL7	100%	100%	100%	100%	99.7%	
MYL9	100%	100%	100%	100%	99.2%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 4, 619365
MYLK	99.2%	99.2%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
MYLK2	100%	100%	100%	100%	99.4%	Cardiomyopathy, hypertrophic, 1, digenic, 192600
MYLK3	100%	100%	100%	100%	99.8%	
MYMK	100%	100%	100%	100%	99.4%	Carey-Fineman-Ziter syndrome, 254940
MYMX	100%	100%	100%	100%	100%	Carey-Fineman-Ziter syndrome 2, 619941
MYO15A	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 3, 600316
MYO18B	100%	100%	100%	99.9%	99.4%	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549
MYO1A	100%	100%	100%	100%	99.6%	?Diarrhea 15, congenital, 621179
MYO1E	100%	100%	100%	100%	99.6%	Glomerulosclerosis, focal segmental, 6, 614131
MYO1H	100%	100%	100%	100%	99.5%	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MYO3A	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 30, 607101;Deafness, autosomal dominant 90, 620722
MYO5A	99%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450
MYO5B	100%	100%	100%	100%	99.5%	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850;Cholestasis, progressive familial intrahepatic, 10, 619868
MYO6	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346;Deafness, autosomal dominant 22, 606346;Deafness, autosomal recessive 37, 607821
MYO7A	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 2, 600060;Usher syndrome, type 1B, 276900;Deafness, autosomal dominant 11, 601317
MYO9A	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYO9B	100%	100%	100%	100%	99.2%	
MYOC	95.3%	95.2%	100%	100%	99.6%	Glaucoma 1A, primary open angle, 137750
MYOCD	100%	100%	100%	99.9%	99.4%	Megabladder, congenital, 618719
MYOD1	100%	100%	100%	99.9%	97.5%	Congenital myopathy 17, 618975
MYOF	98%	98%	100%	100%	99.7%	?Angioedema, hereditary, 7, 619366
MYOM1	100%	100%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MYORG	100%	100%	100%	99.9%	99.1%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
MYOT	100%	100%	100%	100%	99.8%	Myopathy, myofibrillar, 3, 609200
MYOZ2	88.7%	88.7%	100%	100%	99.9%	Cardiomyopathy, hypertrophic, 16, 613838
MYPN	98.4%	98.4%	100%	100%	99.6%	Cardiomyopathy, hypertrophic, 22, 615248;Congenital myopathy 24, 617336;Cardiomyopathy, familial restrictive, 4, 615248;Cardiomyopathy, dilated, 1KK, 615248
MYRF	100%	99.8%	100%	99.9%	99.2%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
MYSM1	100%	100%	100%	100%	99.7%	Bone marrow failure syndrome 4, 618116
MYT1L	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 39, 616521
MYZAP	80%	80%	100%	99.9%	99.7%	Cardiomyopathy, dilated, 2K, 620894
NAA10	100%	100%	99%	89%	68.4%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NAA15	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NAA20	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 73, 619717
NAA60	100%	100%	100%	100%	99.5%	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786
NAA80	100%	100%	100%	99.9%	99.1%	?Auroneurodental syndrome, 620830
NACC1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NADK2	100%	100%	100%	99.9%	99.2%	2,4-dienoyl-CoA reductase deficiency, 616034
NADSYN1	93.5%	93.5%	100%	100%	99.5%	Vertebral, cardiac, renal, and limb defects syndrome 3, 618845
NAE1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210
NAF1	100%	100%	100%	100%	99.5%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NAGA	90%	88%	100%	100%	99.5%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241
NAGLU	100%	100%	100%	100%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NAGS	100%	100%	100%	100%	99.2%	N-acetylglutamate synthase deficiency, 237310
NALCN	100%	100%	100%	100%	99.8%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANOS1	100%	100%	100%	99.4%	96.2%	Spermatogenic failure 12, 615413
NANS	94.8%	94.3%	100%	99.9%	99.7%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NAPB	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 107, 620033
NARS1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092;Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAT8L	100%	98%	100%	99.5%	95.3%	?N-acetylaspartate deficiency, 614063

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NAV3	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with poor or absent speech, dysmorphic facies, and behavioral abnormalities, 621182
NAXD	95.5%	89.7%	100%	100%	99.2%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2, 618321
NAXE	97.6%	93.5%	100%	100%	99.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NBEA	97.6%	97.6%	100%	100%	99.7%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	100%	100%	100%	100%	99.2%	Gray platelet syndrome, 139090
NBN	97.5%	97.5%	100%	100%	99.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCAPD2	97.1%	97.1%	100%	100%	99.7%	Microcephaly 21, primary, autosomal recessive, 617983
NCAPD3	100%	100%	100%	100%	99.7%	Microcephaly 22, primary, autosomal recessive, 617984
NCAPG2	100%	100%	100%	100%	99.8%	Khan-Khan-Katsanis syndrome, 618460

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NCAPH	100%	100%	100%	99.9%	99.4%	?Microcephaly 23, primary, autosomal recessive, 617985
NCDN	99.2%	96.7%	100%	100%	99.4%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCF1	99.7%	96.1%	100%	99.7%	97.8%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCF2	89.5%	89.5%	100%	100%	99.5%	Chronic granulomatous disease 2, autosomal recessive, 233710
NCF4	100%	100%	100%	100%	99.3%	Chronic granulomatous disease 3, autosomal recessive, 613960
NCKAP1	100%	100%	100%	100%	99.7%	
NCKAP1L	100%	100%	100%	100%	99.7%	Immunodeficiency 72 with autoinflammation, 618982
NCOA3	100%	100%	100%	100%	99.8%	
NCOR2	100%	100%	100%	99.9%	98.9%	
NCSTN	100%	100%	100%	100%	99.5%	Acne inversa, familial, 1, 142690
NDC1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with achalasia, polyneuropathy, and alacrima, 621328
NDE1	93.5%	93.5%	100%	100%	99.1%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019
NDN	100%	100%	100%	99.7%	98.1%	
NDNF	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NDP	100%	100%	98.2%	90.3%	76.4%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NDRG1	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4D, 601455
NDST1	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100%	100%	98.8%	90.1%	70.9%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA10	82.9%	80.5%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 22, 618243
NDUFA11	96.1%	95.9%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	80.4%	80.4%	100%	99.9%	99.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA13	99.1%	94.7%	100%	100%	99.5%	{Thyroid carcinoma, Hurthle cell}, 607464;Mitochondrial complex I deficiency, nuclear type 28, 618249
NDUFA2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA3	93.9%	87.2%	100%	100%	98.4%	
NDUFA4	100%	100%	100%	100%	100%	
NDUFA5	75.6%	75.6%	100%	99.9%	99.6%	
NDUFA6	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 33, 618253
NDUFA7	78.2%	78.2%	100%	100%	98.6%	
NDUFA8	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFA9	87%	85.2%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 26, 618247

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NDUFAB1	100%	100%	100%	100%	99.5%	
NDUFAF1	91.6%	91.5%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	90.9%	90.9%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF6	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 17, 618239;Fanconi renotubular syndrome 5, 618913
NDUFAF7	100%	100%	100%	100%	99.9%	
NDUFAF8	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFB1	100%	100%	100%	100%	97.6%	
NDUFB10	100%	100%	100%	100%	99.7%	?Mitochondrial complex I deficiency, nuclear type 35, 619003
NDUFB11	99.9%	99.3%	93.6%	79.3%	59.7%	Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NDUFB2	100%	100%	100%	100%	99.7%	
NDUFB3	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB4	75%	75%	100%	100%	99.5%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NDUFB5	100%	100%	100%	100%	99.7%	
NDUFB6	100%	100%	100%	100%	99.9%	
NDUFB7	72.3%	72.3%	100%	100%	99.7%	?Mitochondrial complex I deficiency, nuclear type 39, 620135
NDUFB8	100%	100%	100%	99.7%	98.9%	Mitochondrial complex I deficiency, nuclear type 32, 618252
NDUFB9	86.8%	86.6%	100%	100%	99.5%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFC1	100%	100%	100%	99.9%	99.2%	
NDUFC2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 36, 619170
NDUFS1	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.9%	98.3%	100%	100%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	98%	92.5%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS5	100%	100%	100%	100%	100%	
NDUFS6	94.7%	86.1%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 2, 618222

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NDUFV1	100%	100%	100%	100%	98.7%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NDUFV3	100%	100%	100%	100%	99.5%	
NEB	99.9%	99.8%	99.6%	99.2%	98.3%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NEBL	100%	100%	100%	100%	99.5%	
NECAP1	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 21, 615833
NECTIN1	93.4%	93.4%	100%	99.9%	99.1%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NECTIN4	100%	100%	100%	100%	99.4%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEDD4L	100%	100%	100%	100%	99.6%	Periventricular nodular heterotopia 7, 617201
NEFH	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924;{?Amyotrophic lateral sclerosis, susceptibility to}, 105400
NEFL	100%	100%	100%	99.9%	98.8%	Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK10	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 44, 618781
NEK11	87.7%	87.7%	100%	100%	99.7%	
NEK2	95.9%	95.9%	100%	100%	100%	?Retinitis pigmentosa 67, 615565
NEK8	100%	100%	100%	100%	99.8%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824
NEK9	100%	100%	100%	100%	99.7%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NEMF	100%	100%	100%	99.8%	99.3%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEPRO	100%	100%	100%	100%	99.5%	
NEU1	100%	100%	100%	99.9%	99.5%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEUROD1	100%	100%	100%	100%	99%	{Type 2 diabetes mellitus, susceptibility to}, 125853;Maturity-onset diabetes of the young 6, 606394

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NEUROD2	100%	100%	100%	99.8%	97%	Developmental and epileptic encephalopathy 72, 618374
NEUROG1	100%	100%	100%	99.6%	97%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469
NEUROG3	100%	100%	100%	99.8%	98.6%	Diarrhea 4, malabsorptive, congenital, 610370
NEXMIF	100%	100%	99.3%	91.3%	72.3%	Intellectual developmental disorder, X-linked 98, 300912
NEXN	100%	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 2M, autosomal recessive, 621261;Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NF2	100%	100%	100%	100%	99.5%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000
NFASC	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFAT5	100%	100%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NFATC1	100%	100%	100%	99.9%	98.5%	
NFATC2	100%	100%	100%	99.9%	99.3%	?Joint contracture, osteochondromas, and B-cell lymphoma, 620232
NFE2	100%	100%	100%	100%	99.7%	
NFE2L2	81.2%	81.2%	100%	100%	99.7%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	92.2%	92.2%	100%	99.9%	99%	Brain malformations with or without urinary tract defects, 613735
NFIB	100%	100%	100%	100%	99.6%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100%	100%	100%	100%	98.8%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NFKB1	100%	100%	100%	100%	99.8%	Immunodeficiency, common variable, 12, 616576
NFKB2	100%	100%	100%	100%	99.5%	Immunodeficiency, common variable, 10, 615577
NFKBIA	100%	100%	100%	100%	99.3%	Ectodermal dysplasia and immunodeficiency 2, 612132
NFS1	83.4%	83.4%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 52, 619386
NFU1	98.9%	94.6%	100%	100%	99.7%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NGF	100%	100%	100%	100%	98.9%	Neuropathy, hereditary sensory and autonomic, type V, 608654
NGLY1	98.4%	97.2%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273
NHEJ1	92.9%	92.9%	100%	100%	99.7%	Microphthalmia/colobo ma 13, 620968;Immunodeficie ncy 124, severe combined, 611291
NHERF1	85.8%	85.8%	100%	100%	99.2%	Nephrolithiasis/osteopo rosis, hypophosphatemic, 2, 612287
NHLH2	100%	100%	100%	100%	99%	?Hypogonadotropic hypogonadism 27 without anosmia, 619755
NHLRC1	100%	100%	100%	99.9%	99.1%	Myoclonic epilepsy of Lafora 2, 620681
NHLRC2	100%	100%	100%	100%	99.8%	FINCA syndrome, 618278
NHP2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 2, 613987
NHS	100%	100%	98.9%	90.1%	71.8%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350
NIN	100%	100%	100%	100%	99.7%	?Seckel syndrome 7, 614851
NIPA1	100%	100%	99.8%	99.2%	97.4%	Spastic paraplegia 6, autosomal dominant, 600363
NIPAL4	100%	100%	100%	99.7%	98.5%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	100%	100%	100%	100%	99.8%	Cornelia de Lange syndrome 1, 122470
NIT1	100%	100%	100%	100%	99.8%	Brain small vessel disease 4, 621313

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NKAP	100%	100%	99.1%	89.5%	71%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NKPD1	100%	100%	100%	99.9%	98.5%	
NKX2-1	100%	100%	100%	99.9%	98.4%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX2-5	100%	100%	100%	99.7%	98.1%	Hypoplastic left heart syndrome 2, 614435;Tetralogy of Fallot, 187500;Hypothyroidism, congenital nongoitrous, 5, 225250;Conotruncal heart malformations, variable, 217095;Ventricular septal defect 3, 614432;Atrial septal defect 7, with or without AV conduction defects, 108900
NKX2-6	100%	100%	100%	100%	99.5%	Persistent truncus arteriosus, 217095;Conotruncal heart malformations, 217095
NKX3-2	100%	100%	100%	100%	99%	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330
NKX6-2	100%	100%	100%	99.6%	97.1%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	100%	100%	100%	99.9%	98.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NLGN3	99.5%	97.9%	97.8%	86.9%	67.8%	{Autism susceptibility, X-linked 1}, 300425
NLGN4X	100%	100%	98.6%	88.6%	69.2%	Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495
NLRC4	100%	100%	100%	100%	99.7%	?Familial cold autoinflammatory syndrome 4, 616115;Autoinflammation with infantile enterocolitis, 616050
NLRP1	98.1%	98.1%	100%	100%	99.7%	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100%	100%	100%	100%	99.3%	Familial cold autoinflammatory syndrome 2, 611762
NLRP14	100%	100%	100%	100%	99.9%	
NLRP2	100%	100%	100%	100%	99.3%	Oocyte/zygote/embryo maturation arrest 18, 620332
NLRP3	100%	100%	100%	100%	99.6%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendotheliitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NLRP5	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 19, 620333
NLRP6	100%	100%	100%	99.8%	98.9%	
NLRP7	100%	100%	100%	100%	99.3%	?Ooxyte/zygote/embryo maturation arrest 25, 621471;Hydatidiform mole, recurrent, 1, 231090
NME1	100%	100%	100%	100%	99.8%	
NME3	100%	100%	100%	100%	99.1%	
NME5	86.6%	86.6%	100%	100%	100%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NME8	100%	100%	100%	100%	99.8%	?Ciliary dyskinesia, primary, 6, 610852
NMNAT1	100%	99.5%	100%	100%	99.4%	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260;Leber congenital amaurosis 9, 608553
NMNAT2	100%	100%	100%	100%	99.7%	
NNT	96.3%	96.3%	100%	100%	99.8%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100%	100%	100%	99.9%	99.1%	Premature ovarian failure 5, 611548
NOD2	100%	100%	100%	100%	99.7%	Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600
NODAL	100%	100%	100%	99.9%	99.8%	Heterotaxy, visceral, 5, autosomal, 270100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NOG	100%	100%	100%	99.7%	98.3%	Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500
NOL3	100%	100%	100%	100%	99.7%	?Myoclonus, familial, 1, 614937
NONO	94.7%	89.1%	99.4%	92.9%	75.5%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOP10	92.5%	92.5%	100%	100%	99.9%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NOP56	100%	98.3%	100%	99.9%	99.5%	Spinocerebellar ataxia 36, 614153
NOS1	100%	100%	100%	100%	99.2%	
NOS1AP	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 22, 619155
NOS2	100%	100%	100%	100%	99.6%	{Malaria, resistance to}, 611162
NOTCH1	99.1%	99%	100%	99.9%	99.4%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NOTCH2	100%	100%	100%	100%	99.8%	Alagille syndrome 2, 610205;Hajdu-Cheney syndrome, 102500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NOTCH2NLC	100%	100%	100%	99.8%	98.4%	Tremor, hereditary essential, 6, 618866;Oculopharyngodistal myopathy 3, 619473;Neuronal intranuclear inclusion disease, 603472
NOTCH3	99.9%	99.4%	100%	100%	99.4%	Lateral meningocele syndrome, 130720;Lipodystrophy, familial partial, type 1, 608600;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 1, 621295;?Myofibromatosis, infantile 2, 615293;Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 1, 125310
NOVA2	100%	100%	99.8%	98.6%	94.2%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPAT	100%	100%	100%	100%	99.8%	
NPC1	100%	100%	100%	100%	99.7%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	99%	95.2%	100%	100%	99.6%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	100%	100%	99.8%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	100%	99.6%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NPHP4	100%	100%	100%	100%	99.5%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
NPHS1	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 1, 256300
NPHS2	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 2, 600995
NPL	91.7%	91.7%	100%	100%	99.7%	
NPM1	87.6%	87.6%	100%	99.9%	99.3%	Leukemia, acute myeloid, somatic, 601626
NPPA	100%	100%	100%	100%	99.7%	Atrial standstill 2, 615745;Atrial fibrillation, familial, 6, 612201
NPPB	100%	100%	100%	100%	100%	
NPPC	100%	100%	100%	100%	99.8%	
NPR2	100%	100%	100%	100%	99.5%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	100%	99.1%	Boudin-Mortier syndrome, 619543
NPRL2	100%	100%	100%	100%	99.6%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	93.8%	93.8%	100%	100%	99.3%	Epilepsy, familial focal, with variable foci 3, 617118
NPSR1	100%	100%	100%	100%	99.7%	?Short sleep, familial natural, 3, 621336
NPTN	85.5%	85.5%	100%	100%	99.4%	
NPTX1	100%	100%	100%	99.9%	98.5%	Spinocerebellar ataxia 50, 620158

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NR0B1	100%	100%	98.6%	88%	68.3%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR0B2	100%	100%	100%	100%	99.5%	Obesity, mild, early-onset, 601665
NR1H4	100%	100%	100%	100%	99.7%	Cholestasis, progressive familial intrahepatic, 5, 617049
NR2E3	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 37, 611131;Enhanced S-cone syndrome 1, 268100
NR2F1	100%	100%	100%	99.9%	98.5%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100%	100%	100%	100%	99.4%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR3C1	100%	100%	100%	100%	99.8%	Glucocorticoid resistance, 615962
NR3C2	100%	100%	100%	99.8%	99.1%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR4A2	100%	100%	100%	99.9%	99.4%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NR4A3	100%	100%	100%	99.8%	98.6%	Chondrosarcoma, extraskeletal myxoid, somatic, 612237

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NR5A1	98.7%	95%	100%	100%	99.5%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
NR6A1	96.3%	96.3%	100%	100%	99.4%	Oculovertebral syndrome, 621277
NRAP	100%	100%	100%	100%	99.7%	
NRAS	89.4%	89.4%	100%	100%	99.9%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NRCAM	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833
NRDC	96.9%	96.9%	100%	100%	99.8%	
NRG1	100%	100%	100%	99.9%	99.3%	
NRIP1	100%	100%	100%	100%	99.8%	?Congenital anomalies of kidney and urinary tract 3, 618270

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NRL	100%	100%	100%	100%	99.4%	Enhanced S-cone syndrome 2, 621371;Retinitis pigmentosa 27, 613750
NRROS	100%	100%	100%	99.9%	99.2%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	100%	100%	99.5%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 621407
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSD2	99.5%	99.5%	100%	100%	99.6%	Rauch-Steindl syndrome, 619695
NSDHL	95.9%	95.9%	99.3%	93.1%	75.4%	CK syndrome, 300831;CHILD syndrome, 308050
NSF	100%	100%	99.8%	98.1%	93.5%	Developmental and epileptic encephalopathy 96, 619340
NSMCE2	92.1%	92.1%	100%	100%	99.9%	Seckel syndrome 10, 617253
NSMCE3	100%	100%	100%	99.9%	98.4%	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241
NSMF	100%	100%	100%	100%	99.2%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NSRP1	91%	91%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001
NSUN2	98.5%	98.5%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 5, 611091

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NSUN3	100%	100%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 48, 619012
NSUN6	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 82, 620779
NT5C2	91.4%	91.4%	100%	100%	99.8%	Spastic paraplegia 45, autosomal recessive, 613162
NT5C3A	100%	100%	100%	100%	99.9%	Anemia, congenital, nonspherocytic hemolytic, 8, 266120
NT5E	91%	91%	100%	100%	99.5%	Calcification of joints and arteries, 211800
NTF4	100%	100%	100%	99.9%	98.8%	Glaucoma 1, open angle, 1O, 613100
NTHL1	100%	100%	100%	99.9%	99.1%	Familial adenomatous polyposis 3, 616415
NTN1	100%	100%	100%	100%	99.1%	Mirror movements 4, 618264
NTNG2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100%	100%	100%	99.9%	99.3%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	89.5%	89.5%	100%	100%	99.5%	Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886
NUAK2	100%	100%	100%	100%	99.5%	?Anencephaly 2, 619452
NUBPL	89.8%	89.8%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDCD3	91%	91%	100%	99.9%	99.2%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NUDT2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with or without peripheral neuropathy, 619844
NUMA1	100%	100%	100%	100%	99.6%	
NUP107	97.8%	97.8%	100%	100%	99.9%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	97.9%	97.9%	100%	100%	99.8%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP155	93.6%	93.6%	100%	100%	99.8%	?Atrial fibrillation 15, 615770
NUP160	94.6%	94.6%	100%	100%	99.8%	?Nephrotic syndrome, type 19, 618178
NUP188	95.4%	95.4%	100%	100%	99.6%	Sandestig-Stefanova syndrome, 618804
NUP205	97%	97%	100%	100%	99.6%	?Nephrotic syndrome, type 13, 616893
NUP210L	100%	100%	100%	100%	99.7%	?Spermatogenic failure 97, 621057
NUP214	100%	100%	100%	100%	99.5%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426
NUP37	100%	100%	100%	100%	100%	?Microcephaly 24, primary, autosomal recessive, 618179
NUP54	100%	100%	100%	100%	99.7%	Dystonia 37, early-onset, with striatal lesions, 620427

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

NUP62	100%	100%	100%	100%	99.3%	Striatonigral degeneration, infantile, 271930
NUP85	100%	99.8%	100%	100%	99.4%	Nephrotic syndrome, type 17, 618176
NUP88	93.5%	93.5%	100%	100%	99.7%	Fetal akinesia deformation sequence 4, 618393
NUP93	95.5%	95.5%	100%	100%	99.7%	Nephrotic syndrome, type 12, 616892
NUS1	93.9%	93.9%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
NUTF2	97.3%	96.9%	100%	99.9%	99.4%	
NUTM2B-AS1						?Oculopharyngeal myopathy with leukoencephalopathy 1, 618637
NXF5						
NXN	100%	100%	100%	99.9%	99%	Robinow syndrome, autosomal recessive 2, 618529
NYNRIN	100%	100%	100%	100%	99.4%	
NYX	100%	100%	98.3%	86.8%	66.3%	Night blindness, congenital stationary (complete), 1A, X-linked, 310500
OAS1	100%	100%	100%	99.9%	99.5%	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042
OAS2	96.6%	96.6%	100%	100%	99.7%	?Autoinflammation and autoimmunity, systemic, with immune dysregulation 2, 621409

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

OAT	100%	100%	100%	99.9%	99.3%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OBSL1	100%	100%	100%	99.8%	99.2%	3-M syndrome 2, 612921
OCA2	93.4%	93.4%	100%	100%	99.7%	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200
OCLN	94.5%	94.5%	100%	99.9%	99.4%	Pseudo-TORCH syndrome 1, 251290
OCRL	100%	100%	99.3%	92.4%	74.8%	Dent disease 2, 300555;Lowe syndrome, 309000
ODAD1	100%	100%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96.1%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 23, 615451
ODAD3	100%	100%	100%	99.9%	98.7%	Ciliary dyskinesia, primary, 30, 616037
ODAD4	100%	100%	100%	99.9%	99.2%	Ciliary dyskinesia, primary, 35, 617092
ODAM	100%	100%	100%	100%	99.5%	
ODAPH	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA4, 614832
ODC1	100%	100%	100%	100%	99.8%	Bachmann-Bupp syndrome, 619075

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

OFD1	100%	100%	99.1%	91%	73.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDH	100%	100%	100%	100%	99.5%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100%	100%	100%	100%	99.5%	
OGG1	100%	100%	100%	100%	99.6%	Renal cell carcinoma, clear cell, somatic, 144700
OGT	100%	100%	99.2%	92%	73.4%	Intellectual developmental disorder, X-linked 106, 300997
OLA1	100%	100%	100%	100%	99.8%	
OOEP	100%	100%	100%	99.9%	99.2%	
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPA3	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPCML	100%	100%	100%	100%	99.6%	Ovarian cancer, somatic, 167000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

OPHN1	94%	94%	98.9%	90.6%	72.9%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
OPLAH	100%	100%	100%	100%	99.3%	5-oxoprolinase deficiency, 260005
OPN1LW	99.5%	99.1%	94.3%	83.2%	66%	Blue cone monochromacy, 303700;Colorblindness, protan, 303900
OPN1MW	99.1%	97.5%	83.4%	70.4%	55.4%	Colorblindness, deutan, 303800;Blue cone monochromacy, 303700
OPN1SW	100%	100%	100%	100%	99.8%	Colorblindness, tritan, 190900
OPTN	100%	100%	100%	100%	99.8%	Glaucoma 1, open angle, E, 137760;Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435;{Glaucoma, normal tension, susceptibility to}, 606657
ORAI1	100%	100%	100%	99.8%	98.6%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
ORC1	100%	100%	100%	100%	99.8%	Meier-Gorlin syndrome 1, 224690
ORC4	90.8%	90.8%	100%	100%	99.7%	Meier-Gorlin syndrome 2, 613800
ORC6	100%	100%	100%	100%	99.7%	Meier-Gorlin syndrome 3, 613803
OSBPL2	100%	100%	100%	100%	99.4%	Dyschromatosis, ichthyosis, deafness, and atopic disease, 621400;Deafness, autosomal dominant 67, 616340

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

OSGEP	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 3, 617729
OSMR	100%	100%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 1, 105250
OSTC	78.3%	78.3%	100%	100%	99.9%	
OSTM1	100%	99.7%	100%	100%	99.5%	Osteopetrosis, autosomal recessive 5, 259720
OTC	100%	100%	99.6%	91.8%	73.3%	Ornithine transcarbamylase deficiency, 311250
OTOA	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 22, 607039
OTOF	100%	100%	100%	100%	99.3%	Auditory neuropathy, autosomal recessive, 1, 601071;Deafness, autosomal recessive 9, 601071
OTOG	100%	100%	100%	99.9%	99.3%	Deafness, autosomal recessive 18B, 614945
OTOGL	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 84B, 614944
OTUD5	100%	100%	97.7%	84.3%	65.5%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	99.9%	99.1%	100%	99.9%	97.8%	Neurodevelopmental disorder with hypotonia and seizures, 620790

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

OTULIN	100%	100%	100%	100%	99.4%	Autoinflammation, panniculitis, and dermatosis syndrome, autosomal recessive, 617099;{Immunodeficiency 107, susceptibility to invasive staphylococcus aureus infection}, 619986;Autoinflammation, panniculitis, and dermatosis syndrome, autosomal dominant, 621030
OTX2	100%	100%	100%	100%	99.9%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OVOL2	100%	100%	100%	99.9%	99.2%	Corneal dystrophy, posterior polymorphous, 1, 122000
OXA1L	100%	100%	100%	100%	99.7%	
OXCT1	90.5%	90.5%	100%	100%	99.7%	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050
OXGR1	100%	100%	100%	100%	99.8%	Nephrolithiasis, calcium oxalate, 2, with nephrocalcinosis, 620374
OXR1	100%	100%	100%	100%	99.8%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P2RX2	100%	100%	100%	99.9%	98.4%	Deafness, autosomal dominant 41, 608224
P2RY12	100%	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 8, 609821

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

P3H1	95.3%	93.2%	100%	99.9%	99.1%	Osteogenesis imperfecta, type VIII, 610915
P3H2	100%	100%	100%	99.8%	99.2%	Myopia, high, with cataract and vitreoretinal degeneration, 614292
P4HA2	100%	100%	100%	100%	99.6%	Myopia 25, autosomal dominant, 617238
P4HB	90.9%	89.4%	100%	99.9%	99%	Cole-Carpenter syndrome 1, 112240
P4HTM	89.4%	89.4%	100%	99.9%	99.2%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPC1	94.3%	93.8%	100%	100%	99.7%	
PABPC1L	100%	100%	100%	99.9%	99.5%	Oocyte/zygote/embryo maturation arrest 22, 621093
PABPN1	100%	100%	100%	100%	98.8%	Oculopharyngeal muscular dystrophy, 164300
PACS1	100%	100%	100%	100%	99.3%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	78.8%	78.8%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 66, 618067
PACSIN3	100%	100%	100%	100%	99.7%	Congenital myopathy 27, 621343
PADI3	100%	100%	100%	100%	99.6%	Uncombable hair syndrome, 191480
PADI6	100%	100%	100%	99.9%	99.1%	Oocyte/zygote/embryo maturation arrest 16, 617234
PAFAH1B1	100%	100%	100%	100%	99.8%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PAH	94.9%	93.7%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PAICS	100%	100%	100%	100%	99.8%	?Phosphoribosylaminoimidazole carboxylase deficiency, 619859
PAK1	100%	100%	100%	100%	99.9%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK2	100%	100%	100%	100%	99.8%	?Knobloch syndrome 2, 618458
PAK3	100%	100%	99.3%	91.9%	73.7%	Intellectual developmental disorder, X-linked 30, 300558
PALB2	100%	100%	100%	100%	99.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PALS1	100%	100%	100%	100%	99.7%	
PAM16	85.8%	85.5%	100%	100%	99.3%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Me like type, 613320
PAN2	100%	100%	100%	100%	99.7%	Developmental delay with variable cardiac and renal congenital anomalies and dysmorphic facies, 621384
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200
PANK4	100%	100%	100%	100%	99.1%	?Cataract 49, 619593

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PANX1	100%	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 7, 618550
PAPPA2	100%	100%	100%	100%	99.7%	Short stature, Dauber-Argente type, 619489
PAPSS2	100%	100%	100%	100%	99.6%	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847
PARK7	97.5%	93.1%	100%	99.8%	99.4%	Parkinson disease 7, autosomal recessive early-onset, 606324
PARN	97.2%	96%	100%	100%	99.8%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP4	100%	100%	100%	100%	99.7%	
PARP6	100%	100%	100%	100%	99.6%	
PARS2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 75, 618437
PATL2	100%	100%	100%	100%	99.8%	Oocyte/zygote/embryo maturation arrest 4, 617743
PAX1	100%	100%	100%	100%	98.6%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PAX2	100%	100%	100%	100%	99.2%	Glomerulosclerosis, focal segmental, 7, 616002;Papillorenal syndrome, 120330

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PAX3	100%	100%	100%	100%	99.6%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PAX4	100%	100%	100%	99.9%	99.4%	{Diabetes mellitus, ketosis-prone, susceptibility to}, 612227;Maturity-onset diabetes of the young, type IX, 612225;Diabetes mellitus, type 2, 125853
PAX5	100%	100%	100%	99.9%	99.5%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PAX6	100%	100%	100%	100%	99.5%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia/coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190
PAX7	94.2%	94.2%	100%	99.9%	99.1%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100%	100%	100%	99.9%	99.5%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PAX9	100%	100%	100%	100%	99.4%	Tooth agenesis, selective, 3, 604625
PBRM1	100%	100%	100%	100%	99.8%	?Renal cell carcinoma, clear cell, 144700
PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	99.9%	99.2%	100%	100%	99.5%	Pyruvate carboxylase deficiency, 266150
PCARE	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 54, 613428
PCBD1	100%	100%	100%	100%	99.5%	Hyperphenylalaninemia, BH4-deficient, D, 264070
PCCA	84.6%	84.4%	100%	100%	99.7%	Propionicacidemia, 606054
PCCB	86.8%	85.3%	100%	100%	99.8%	Propionicacidemia, 606054
PCDH12	100%	100%	100%	100%	99.5%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH15	100%	100%	100%	100%	99.6%	Usher syndrome, type 1D/F digenic, 601067;Deafness, autosomal recessive 23, 609533;Usher syndrome, type 1F, 602083
PCDH19	100%	100%	98.2%	86.7%	65.6%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880
PCGF2	100%	100%	100%	100%	98.8%	Turnpenny-Fry syndrome, 618371

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PCK1	100%	100%	100%	100%	99.7%	Phosphoenolpyruvate carboxykinase deficiency, cytosolic, 261680
PCK2	100%	100%	100%	100%	99.5%	PEPCK deficiency, mitochondrial, 261650
PCLO	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 3, 608027
PCNA	100%	100%	100%	100%	99.8%	?Ataxia-telangiectasia-like disorder 2, 615919
PCNT	100%	100%	100%	100%	99.6%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCSK1	100%	100%	100%	100%	99.9%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PCSK9	95%	95%	100%	100%	99.5%	{Low density lipoprotein cholesterol level QTL 1}, 603776;Hypercholesterolemia, familial, 3, 603776
PCYT1A	100%	100%	100%	100%	99.7%	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940;Lipodystrophy, congenital generalized, type 5, 620680
PCYT2	99.9%	97.1%	100%	100%	99.5%	Spastic paraplegia 82, autosomal recessive, 618770
PDCD1	100%	100%	100%	99.9%	98.9%	?Autoimmune disease, multisystem, infantile-onset, 4, 621004
PDCD10	100%	100%	100%	100%	100%	Cerebral cavernous malformations-3, 603285

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PDCD6IP	100%	100%	100%	100%	99.8%	?Microcephaly 29, primary, autosomal recessive, 620047
PDE10A	100%	99.4%	99.9%	99.7%	98.3%	Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE11A	100%	100%	100%	100%	99.7%	Pigmented nodular adrenocortical disease, primary, 2, 610475
PDE12	93%	93%	100%	100%	99.5%	
PDE1C	100%	100%	100%	100%	99.8%	?Deafness, autosomal dominant 74, 618140
PDE2A	100%	100%	100%	100%	99.3%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE3A	100%	100%	100%	100%	99.5%	Hypertension and brachydactyly syndrome, 112410
PDE4D	100%	100%	100%	100%	99.2%	Acrodysostosis 2, with or without hormone resistance, 614613
PDE6A	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 43, 613810
PDE6B	100%	100%	100%	100%	99.2%	Retinitis pigmentosa-40, 613801;Night blindness, congenital stationary, autosomal dominant 2, 163500
PDE6C	100%	100%	100%	100%	99.9%	Cone dystrophy 4, 613093
PDE6D	100%	100%	100%	100%	99.1%	Joubert syndrome 22, 615665
PDE6G	100%	100%	100%	100%	99.2%	Retinitis pigmentosa 57, 613582
PDE6H	100%	100%	100%	100%	99.9%	Achromatopsia 6, 610024

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PDE8B	100%	100%	100%	99.9%	99.4%	Pigmented nodular adrenocortical disease, primary, 3, 614190;Striatal degeneration, autosomal dominant, 609161
PDGFB	99.7%	94.7%	100%	100%	99.8%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483
PDGFRA	100%	100%	100%	100%	99.6%	Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial, 175510;Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685
PDGFRB	99.3%	97.4%	100%	100%	99.2%	Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PDGFRL	92.4%	92.4%	100%	100%	99.8%	Hepatocellular cancer, somatic, 114550;Colorectal cancer, somatic, 114500
PDHA1	91.4%	89.5%	99.3%	91.4%	72.9%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHA2	100%	100%	100%	100%	99.6%	Spermatogenic failure 70, 619828
PDHB	100%	100%	100%	100%	99.8%	Pyruvate dehydrogenase E1-beta deficiency, 614111

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PDHX	91.6%	91.6%	100%	100%	99.6%	Lacticacidemia due to PDX1 deficiency, 245349
PDIA6	100%	100%	100%	100%	99.5%	
PDK1	100%	100%	100%	100%	99.4%	
PDK2	92%	92%	100%	100%	99.3%	
PDK3	100%	100%	99.1%	91%	73.3%	?Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905
PDK4	100%	100%	100%	100%	99.7%	
PDLIM3	100%	100%	100%	100%	99.6%	
PDLIM5	99.9%	98.2%	100%	100%	99.8%	
PDP1	100%	100%	100%	100%	99.9%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PDX1	100%	100%	100%	100%	99.5%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PDXK	91.6%	88.3%	100%	100%	99.3%	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy, 618511
PDYN	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia 23, 610245

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PDZD7	100%	99.8%	100%	100%	99.3%	Deafness, autosomal recessive 57, 618003;{Retinal disease in Usher syndrome type IIA, modifier of}, 276901;Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PDZD8	100%	100%	100%	99.8%	98.5%	Intellectual developmental disorder with autism and dysmorphic facies, 620021
PEPD	94%	94%	100%	100%	99.6%	Prolidase deficiency, 170100
PER2	100%	100%	100%	100%	99.7%	?Advanced sleep phase syndrome, familial, 1, 604348
PER3	100%	100%	100%	100%	99.5%	?Advanced sleep phase syndrome, familial, 3, 616882
PERCC1	100%	100%	100%	100%	99.8%	Diarrhea 11, malabsorptive, congenital, 618662
PERP	100%	100%	100%	100%	99.4%	Erythrokeratodermia variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208
PET100	100%	100%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PET117	100%	100%	100%	100%	99.5%	?Mitochondrial complex IV deficiency, nuclear type 19, 619063
PEX1	100%	100%	100%	100%	99.9%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	92.4%	92.4%	100%	100%	99.9%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	88.6%	88.6%	100%	100%	99.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	91.9%	91.7%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PEX3	100%	100%	100%	100%	99.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.8%	99%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PFAS	100%	100%	100%	100%	99.3%	
PFKM	100%	100%	100%	100%	99.7%	Glycogen storage disease VII, 232800
PFN1	100%	100%	100%	100%	99.5%	Amyotrophic lateral sclerosis 18, 614808
PGAM2	100%	100%	100%	99.8%	99.4%	Glycogen storage disease X, 261670
PGAP1	96.7%	96.7%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100%	100%	100%	99.9%	98.9%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PGAP3	100%	100%	100%	100%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGBD5	100%	100%	100%	99.9%	99.4%	Neurodevelopmental disorder with seizures, hypotonia, and variable spasticity, 621482
PGK1	92.7%	92.7%	99.3%	90.9%	71.5%	Phosphoglycerate kinase 1 deficiency, 300653
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PGM2L1	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100%	100%	100%	100%	99.8%	Immunodeficiency 23, 615816
PHACTR1	100%	100%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 70, 618298
PHC1	100%	100%	100%	99.9%	99.4%	?Microcephaly 11, primary, autosomal recessive, 615414
PHEX	100%	100%	99%	92.2%	75%	Hypophosphatemic rickets, X-linked dominant, 307800
PHF21A	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF5A	85%	85%	100%	100%	99.8%	
PHF6	100%	100%	99.4%	92.6%	74.6%	Borjeson-Forssman-Lehmann syndrome, 301900

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PHF8	100%	100%	98.9%	90.4%	71.6%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263
PHGDH	100%	100%	100%	100%	99.6%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	99.4%	99.3%	100%	99.9%	99.3%	Chung-Jansen syndrome, 617991
PHKA1	100%	100%	99.5%	91.9%	72.6%	Muscle glycogenosis, 300559
PHKA2	100%	100%	99.1%	90.1%	71%	Glycogen storage disease, type IXa2, 306000;Glycogen storage disease, type IXa1, 306000
PHKB	100%	100%	100%	100%	99.7%	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750
PHKG1	97.4%	92.5%	100%	100%	99.3%	
PHKG2	100%	100%	100%	100%	99.1%	Glycogen storage disease IXc, 613027
PHLDB1	97.7%	97.5%	100%	99.9%	99.1%	Osteogenesis imperfecta, type XXIII, 620639
PHOX2A	100%	100%	100%	99.9%	99.2%	Fibrosis of extraocular muscles, congenital, 2, 602078
PHOX2B	100%	100%	100%	100%	98.6%	{Neuroblastoma, susceptibility to, 2}, 613013;Neuroblastoma with Hirschsprung disease, 613013;Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PI4K2A	86.9%	86.9%	100%	100%	99.1%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732
PI4KA	99.3%	99.2%	100%	99.9%	99%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PI4KB	100%	100%	100%	100%	99.3%	Deafness, autosomal dominant 87, 620281
PIBF1	100%	100%	100%	100%	99.8%	Joubert syndrome 33, 617767
PICALM	100%	100%	100%	100%	99.8%	Leukemia, acute myeloid, somatic, 601626
PICK1	100%	100%	100%	100%	99.5%	
PIDD1	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827
PIEZO1	100%	100%	100%	100%	99.2%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIGA	100%	100%	99.2%	90.8%	73.4%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100%	100%	100%	100%	99.8%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356
PIGG	100%	100%	100%	100%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	82.9%	76.2%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIGK	88.2%	88.2%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	100%	100%	99.7%	CHIME syndrome, 280000
PIGM	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol deficiency, 610293
PIGN	95.9%	94.4%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	100%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	100%	99.6%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	98.4%	95.1%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	100%	100%	99.5%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	97.1%	97.1%	100%	100%	99.7%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIGV	100%	100%	100%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIGW	100%	100%	100%	100%	99.9%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	100%	100%	99.8%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809
PIK3C2A	100%	100%	100%	100%	99.9%	Oculoskeletodental syndrome, 618440

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIK3CA	100%	100%	100%	100%	99.9%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3CD	100%	100%	100%	100%	99.3%	Immunodeficiency 14A, autosomal dominant, 615513;Immunodeficiency 14B, autosomal recessive, 619281;?Roifman-Chitayat syndrome, digenic, 613328
PIK3CG	100%	100%	100%	100%	99.7%	Immunodeficiency 97 with autoinflammation, 619802

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PIK3R1	100%	100%	100%	100%	99.7%	Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	100%	100%	100%	100%	99.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIK3R5	100%	100%	100%	100%	98.9%	Ataxia-oculomotor apraxia 3, 615217
PIKFYVE	100%	100%	100%	100%	99.8%	Corneal fleck dystrophy, 121850
PINK1	96.9%	96.9%	100%	100%	99.2%	Parkinson disease 6, early onset, 605909
PIP5K1C	90.6%	90.2%	100%	99.9%	99.4%	Lethal congenital contractural syndrome 3, 611369
PISD	100%	100%	100%	99.9%	99.5%	Liberfarb syndrome, 618889
PITPNM3	100%	100%	100%	100%	98.7%	Cone-rod dystrophy 5, 600977
PITRM1	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PITX1	100%	100%	100%	100%	98.8%	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800
PITX2	100%	100%	100%	100%	99.1%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PITX3	100%	100%	100%	99.9%	99.3%	Cataract 11, multiple types, 610623;Anterior segment dysgenesis 1, multiple subtypes, 107250;Cataract 11, syndromic, autosomal recessive, 610623
PIWIL2	98.5%	98.5%	100%	100%	99.7%	
PJA1	100%	100%	98.3%	86%	67.4%	
PJVK	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 59, 610220
PKD1	100%	99.9%	100%	99.9%	99.1%	Polycystic kidney disease 1, 173900
PKD1L1	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	100%	100%	100%	100%	99.1%	Polycystic kidney disease 2, 613095
PKDCC	100%	100%	100%	99.9%	98.2%	Rhizomelic limb shortening with dysmorphic features, 618821
PKHD1	100%	100%	100%	100%	99.7%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PKHD1L1	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 124, 620794
PKLR	100%	100%	100%	100%	99.4%	Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient, 266200;[Adenosine triphosphate, elevated, of erythrocytes], 102900
PKP1	100%	100%	100%	100%	99.6%	Ectodermal dysplasia/skin fragility syndrome, 604536
PKP2	98.9%	98.1%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 9, 609040

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PLA2G4A	100%	100%	100%	100%	99.8%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G5	100%	100%	100%	100%	99.3%	[Fleck retina, familial benign], 228980
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLA2G7	100%	100%	100%	100%	99.6%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAA	100%	100%	100%	99.9%	99.8%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAAT3	65.6%	63.1%	100%	100%	99.7%	Lipodystrophy, familial partial, type 9, 620683
PLAG1	100%	100%	100%	100%	99.8%	Adenomas, salivary gland pleomorphic, somatic, 181030;Silver-Russell syndrome 4, 618907
PLAT	96.1%	96.1%	100%	100%	99.4%	
PLAU	100%	100%	100%	100%	99.4%	Quebec platelet disorder, 601709;{Alzheimer disease, late-onset, susceptibility to}, 104300
PLCB1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 12, 613722
PLCB3	100%	100%	100%	100%	99.6%	Spondylometaphyseal dysplasia with corneal dystrophy, 618961

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PLCB4	99%	99%	100%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
PLCD1	100%	100%	100%	100%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCE1	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 3, 610725
PLCG1	100%	100%	100%	100%	99.6%	?Immune dysregulation, autoimmunity, and autoinflammation, 620514
PLCG2	96.3%	96.3%	100%	100%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
PLCH1	100%	100%	100%	100%	99.8%	Holoprosencephaly 14, 619895
PLCZ1	100%	100%	100%	99.7%	98.6%	Spermatogenic failure 17, 617214
PLD1	100%	100%	100%	100%	99.8%	Cardiac valvular dysplasia 1, 212093
PLD3	100%	100%	100%	100%	99.3%	?Spinocerebellar ataxia 46, 617770
PLD4	100%	100%	100%	100%	99.7%	Systemic lupus erythematosus 18, 621369
PLD6	100%	100%	100%	99.8%	98.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PLEC	100%	100%	100%	100%	98.9%	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487;Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138;Epidermolysis bullosa simplex 5A, Ogna type, 131950;Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLEKHG2	100%	100%	100%	100%	99.4%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLEKHG5	100%	100%	100%	99.9%	99.3%	Neuronopathy, distal hereditary motor, autosomal recessive 4, 611067;Charcot-Marie-Tooth disease, recessive intermediate C, 615376
PLEKHM1	100%	100%	100%	99.8%	98.8%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PLEKHM2	96.7%	96.7%	100%	100%	99.5%	
PLG	89.3%	89.3%	100%	100%	99.8%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PLIN1	100%	100%	100%	99.9%	99.1%	Lipodystrophy, familial partial, type 4, 613877

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PLIN4	99.9%	99.9%	99.8%	99.4%	97.6%	Myopathy with rimmed ubiquitin-positive autophagic vacuolation, autosomal dominant, 601846
PLK1	100%	100%	100%	100%	99.3%	
PLK4	100%	100%	100%	100%	99.9%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLN	100%	100%	100%	100%	99.9%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PLOD1	96.5%	96.5%	100%	100%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	100%	100%	100%	99.9%	99.4%	Bruck syndrome 2, 609220
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PLP1	94.5%	94.1%	98.8%	88.7%	69.7%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PLPBP	100%	100%	100%	99.8%	98.6%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PLPP6	100%	100%	100%	100%	99.1%	
PLS1	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 76, 618787
PLS3	94.2%	94.2%	99.4%	91.1%	72.5%	Bone mineral density QTL18, osteoporosis, 300910;Diaphragmatic hernia 5, X-linked, 306950

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PLVAP	94.1%	94.1%	100%	99.8%	98.6%	Diarrhea 10, protein-losing enteropathy type, 618183
PLXNA1	100%	100%	100%	100%	99.6%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PLXNA2	100%	100%	100%	100%	99.4%	
PLXNB2	100%	100%	100%	100%	99.1%	
PLXND1	100%	100%	100%	99.9%	99.2%	Congenital heart defects, multiple types, 9, 620294
PMEPA1	100%	100%	100%	100%	98.9%	
PMFBP1	100%	100%	100%	100%	99.6%	Spermatogenic failure 31, 618112
PML	96.6%	96.6%	100%	100%	99.5%	
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP2	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, demyelinating, type 1G, 618279
PMP22	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 1A, 118220;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Neuropathy, recurrent, with pressure palsies, 162500;Dejerine-Sottas disease, 145900
PMPCA	96.2%	96.2%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100%	100%	99.8%	Multiple mitochondrial dysfunctions syndrome 6, 617954

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PMS2	93.4%	93.4%	99.8%	99.6%	98.9%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PMS2CL						
PMVK	71.2%	71.2%	100%	100%	99.5%	Porokeratosis 1, multiple types, 175800
PNKD	100%	100%	100%	100%	99.4%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100%	100%	100%	99.9%	99.2%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNLDC1	95.2%	95.2%	100%	100%	99.6%	Spermatogenic failure 57, 619528
PNLIP	100%	100%	100%	100%	99.6%	?Pancreatic lipase deficiency, 614338
PNMT	100%	100%	100%	99.9%	99.3%	
PNP	100%	100%	100%	100%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA1	100%	100%	100%	99.9%	98.9%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	80.4%	80.4%	100%	100%	99.6%	Neutral lipid storage disease with myopathy, 610717
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PNPLA8	100%	100%	100%	100%	99.5%	Mitochondrial myopathy with lactic acidosis, 251950
PNPO	100%	100%	100%	100%	99.6%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POC1A	100%	100%	100%	99.8%	99.1%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POC1B	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 20, 615973
POC5	93.4%	93.4%	100%	100%	99.9%	
PODXL	91.7%	91.4%	100%	100%	99.3%	
POF1B	95.4%	95.4%	99.5%	92.7%	74.6%	?Premature ovarian failure 2B, 300604
POFUT1	100%	100%	100%	99.9%	99.6%	Dowling-Degos disease 2, 615327
POGLUT1	99.9%	98%	100%	100%	99.8%	Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POGZ	100%	100%	100%	100%	99.6%	White-Sutton syndrome, 616364
POLA1	100%	100%	99.3%	91.7%	73.3%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POLD1	100%	100%	100%	99.9%	99%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLD3	100%	100%	100%	100%	99.5%	Immunodeficiency 122, 620869
POLE	100%	100%	100%	100%	99.3%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLE2	100%	100%	100%	100%	99.9%	
POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POLG2	95.8%	95.8%	100%	100%	99.5%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131;?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528;?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425
POLH	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, variant type, 278750
POLL	99%	96.3%	100%	99.9%	99.4%	
POLR1A	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1B	100%	100%	100%	100%	99.7%	Treacher-Collins syndrome 4, 618939
POLR1C	83.6%	83.5%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717
POLR2A	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3F	91.4%	90.9%	100%	100%	99.9%	?Immunodeficiency 101 (varicella zoster virus-specific), 619872
POLR3GL	100%	100%	100%	100%	99.5%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POLR3K	100%	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	99.7%	98.7%	100%	100%	99%	Combined oxidative phosphorylation deficiency 55, 619743
POMC	100%	100%	100%	100%	99.7%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMGNT1	100%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151;Retinitis pigmentosa 76, 617123;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POMGNT2	100%	100%	100%	100%	99.5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100%	100%	100%	100%	99.8%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMP	83.2%	83.2%	100%	100%	99.6%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
POMT1	100%	99.7%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POMT2	94.9%	94.9%	100%	99.8%	98.4%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156
POP1	100%	100%	100%	100%	99.6%	Anauxetic dysplasia 2, 617396
POPDC1	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 25, 616812
POPDC2	100%	100%	100%	100%	99.7%	Cardiac conduction disease with or without cardiomyopathy 2, 621367
POPDC3	100%	100%	100%	100%	99.6%	Muscular dystrophy, limb-girdle, autosomal recessive 26, 618848
POR	98%	94.9%	100%	100%	99.6%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	100%	100%	98.5%	89.4%	70.4%	Focal dermal hypoplasia, 305600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

POT1	100%	100%	100%	99.9%	99.3%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
POU1F1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU2AF1	100%	100%	100%	100%	99.6%	
POU3F2	100%	100%	100%	99.9%	98.7%	
POU3F3	100%	99.8%	99.9%	97.9%	88.4%	Snijders Blok-Fisher syndrome, 618604
POU3F4	100%	100%	98%	86%	66.8%	Deafness, X-linked 2, 304400
POU4F1	95.7%	92.7%	100%	99.5%	96.9%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
POU4F3	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 15/52, 602459
POU6F2	100%	100%	100%	99.9%	99.2%	{Wilms tumor susceptibility-5}, 601583
PPA2	82.6%	82.6%	100%	100%	99.7%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PPARG	95.8%	95.8%	100%	100%	99.8%	{Diabetes, type 2}, 125853;Insulin resistance, severe, digenic, 125853;Lipodystrophy, familial partial, type 3, 604367;Obesity, severe, 601665

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PPCDC	100%	100%	100%	100%	100%	
PPCS	100%	100%	100%	100%	99.3%	Cardiomyopathy, dilated, 2C, 618189
PPFIA2	100%	100%	100%	100%	99.8%	
PPFIA3	100%	100%	100%	99.8%	98.5%	Paul-Chao neurodevelopmental syndrome, 621122
PPFIBP1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPIB	100%	100%	100%	100%	99.6%	Osteogenesis imperfecta, type IX, 259440
PPIL1	80.1%	80.1%	100%	100%	99.2%	Pontocerebellar hypoplasia, type 14, 619301
PPIP5K2	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 100, 618422
PPM1D	100%	100%	100%	100%	99.9%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPM1K	100%	100%	100%	100%	99.8%	Maple syrup urine disease, mild variant, 615135
PPOX	99.9%	97.6%	100%	100%	99.3%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PPP1CB	87.8%	87.6%	100%	100%	99.7%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	100%	100%	100%	100%	99.8%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R13L	100%	100%	100%	99.9%	98.5%	Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities, 620519

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PPP1R15B	100%	100%	100%	100%	99.8%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP1R3A	100%	100%	100%	100%	99.7%	Insulin resistance, severe, digenic, 125853
PPP1R3F	97%	97%	98.6%	88.8%	70.5%	
PPP2CA	100%	100%	100%	100%	99.8%	Houge-Janssens syndrome 3, 618354
PPP2R1A	94.1%	94%	100%	99.9%	99.5%	Houge-Janssens syndrome 2, 616362
PPP2R1B	100%	100%	100%	100%	99.8%	Lung cancer, somatic, 211980
PPP2R2B	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 12, 604326
PPP2R3C	100%	100%	100%	100%	99.9%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PPP2R5B	100%	100%	100%	100%	99.6%	
PPP2R5C	97.8%	96.9%	100%	100%	99.7%	Houge-Janssens syndrome 4, 621185
PPP2R5D	100%	100%	100%	100%	99.3%	Houge-Janssens syndrome 1, 616355
PPP3CA	100%	100%	100%	100%	99.6%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PPT1	90.3%	90.3%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PQBP1	100%	100%	98.6%	86.7%	67.3%	Renpenning syndrome, 309500
PRCC	92.2%	92.2%	100%	100%	99.4%	Renal cell carcinoma, papillary, 605074
PRCD	100%	100%	100%	99.8%	99.7%	Retinitis pigmentosa 36, 610599
PRDM10	100%	100%	100%	100%	99.4%	?Birt-Hogg-Dube syndrome 2, 620459
PRDM12	97.1%	92.6%	100%	99.9%	98.8%	Neuropathy, hereditary sensory and autonomic, type VIII, 616488
PRDM13	100%	100%	100%	99.9%	98.7%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM15	100%	100%	100%	99.9%	99.2%	
PRDM16	100%	100%	100%	99.9%	98.9%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373
PRDM5	100%	100%	100%	100%	99.8%	Brittle cornea syndrome 2, 614170
PRDM6	100%	100%	100%	99.9%	98.8%	Patent ductus arteriosus 3, 617039
PRDM8	100%	100%	100%	99.8%	97.5%	?Epilepsy, progressive myoclonic, 10, 616640
PRDX1	100%	100%	100%	100%	99.3%	Methylmalonic aciduria and homocystinuria, cblC type, digenic, 277400
PRDX2	100%	100%	100%	100%	99.6%	
PRDX3	100%	98.8%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PREPL	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 22, 616224
PRF1	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRG4	100%	100%	100%	99.8%	98.4%	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250
PRICKLE1	100%	100%	100%	100%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	100%	100%	100%	100%	99.5%	
PRIM1	91.8%	91.8%	100%	100%	99.7%	Primordial dwarfism-immunodeficiency-lipodystrophy syndrome, 620005
PRIMPOL	93.5%	93.5%	100%	100%	99.7%	Myopia 22, autosomal dominant, 615420
PRKAA1	100%	100%	100%	100%	99.8%	
PRKACA	100%	100%	100%	100%	99.4%	Cushing syndrome, ACTH-independent adrenal, somatic, 615830;Cardioacrofacial dysplasia 1, 619142
PRKACB	94.2%	94.2%	100%	99.9%	99.6%	Cardioacrofacial dysplasia 2, 619143
PRKACG	100%	100%	100%	99.6%	98.3%	?Bleeding disorder, platelet-type, 19, 616176
PRKAG2	100%	100%	100%	99.9%	99.6%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PRKAR1A	100%	100%	100%	100%	99.6%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960
PRKAR1B	100%	100%	100%	99.9%	99.2%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRKCA	100%	100%	100%	100%	99.5%	Pituitary tumor, invasive
PRKCB	96.4%	96.4%	100%	100%	99.7%	
PRKCD	100%	100%	100%	100%	99.7%	Autoimmune lymphoproliferative syndrome, type III, 615559
PRKCG	100%	100%	100%	99.8%	98.3%	Spinocerebellar ataxia 14, 605361
PRKCSH	100%	99.6%	100%	99.9%	99.4%	Polycystic liver disease 1, 174050
PRKCZ	100%	100%	100%	100%	99.1%	
PRKD1	100%	100%	100%	99.8%	98.9%	Congenital heart defects and ectodermal dysplasia, 617364
PRKDC	100%	100%	100%	100%	99.7%	Immunodeficiency 26, with or without neurologic abnormalities, 615966
PRKG1	95.9%	95.9%	100%	99.9%	99.6%	Aortic aneurysm, familial thoracic 8, 615436
PRKG2	87.9%	87.9%	100%	100%	99.8%	Acromesomelic dysplasia 4, 619636;?Spondylometaphyseal dysplasia, Pagnamenta type, 619638

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PRKN	100%	100%	100%	100%	99.8%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	100%	99.8%	Dystonia 16, 612067
PRLR	100%	100%	100%	100%	99.8%	Multiple fibroadenomas of the breast, 615554;Hyperprolactinemia, 615555
PRMT7	100%	100%	100%	100%	99.6%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRMT9	97.7%	97.7%	99.9%	98.9%	96.2%	
PRNP	100%	100%	100%	100%	99.8%	Spongiform encephalopathy with neuropsychiatric features, 606688;Gerstmann-Straussler disease, 137440;Huntington disease-like 1, 603218;Insomnia, fatal familial, 600072;{Kuru, susceptibility to}, 245300;Cerebral amyloid angiopathy, PRNP-related, 137440;Creutzfeldt-Jakob disease, 123400
PROC	100%	100%	100%	100%	99.6%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860;Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PRODH	91.9%	91.9%	100%	99.9%	99.5%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PROK2	100%	100%	100%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROM1	100%	100%	100%	100%	99.7%	Macular dystrophy, retinal, 2, 608051;Retinitis pigmentosa 41, 612095;Stargardt disease 4, 603786;Cone-rod dystrophy 12, 612657
PROP1	100%	100%	100%	99.9%	99.1%	Pituitary hormone deficiency, combined, 2, 262600
PRORP	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 54, 619737
PROS1	100%	100%	100%	100%	99.7%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514;Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PROZ	97.7%	94.5%	100%	100%	99.1%	[Protein Z deficiency], 614024
PRPF19	100%	100%	100%	100%	99.4%	
PRPF3	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 18, 601414
PRPF31	93.1%	90.1%	100%	100%	99.4%	Retinitis pigmentosa 11, 600138
PRPF4	90.5%	87.4%	100%	100%	99.6%	Retinitis pigmentosa 70, 615922
PRPF6	96.2%	96.2%	100%	100%	99.4%	Retinitis pigmentosa 60, 613983
PRPF8	100%	100%	100%	100%	99.3%	Retinitis pigmentosa 13, 600059

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PRPH2	100%	100%	100%	100%	99.4%	Macular dystrophy, patterned, 1, 169150;Choroidal dystrophy, central areolar 2, 613105;Retinitis punctata albescens, 136880;Leber congenital amaurosis 18, 608133;Macular dystrophy, vitelliform, 3, 608161;Retinitis pigmentosa 7 and digenic form, 608133
PRPS1	100%	100%	99%	89.8%	71.8%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PRR11	100%	100%	100%	99.9%	99.3%	
PRR12	100%	100%	100%	99.9%	98.7%	Neuroocular syndrome, 619539
PRRT2	100%	100%	100%	100%	98.5%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200
PRRX1	100%	100%	100%	100%	99.5%	Agnathia-otocephaly complex, 202650
PRSS1	100%	100%	100%	99.6%	96.8%	Pancreatitis, hereditary, 167800
PRSS12	100%	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 1, 249500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PRSS56	100%	100%	100%	100%	99.5%	Microphthalmia, isolated 6, 613517
PRUNE1	93.1%	93.1%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PRX	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4F, 614895;Dejerine-Sottas disease, 145900
PSAP	99.6%	97.1%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PSAT1	100%	100%	100%	100%	99.9%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSEN1	100%	99.9%	100%	100%	99.7%	Pick disease, 172700;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;?Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with or without spastic paraparesis, 607822
PSEN2	70.1%	70%	100%	99.9%	99.3%	Alzheimer disease-4, 606889;Cardiomyopathy, dilated, 1V, 613697
PSENEN	100%	100%	100%	100%	99.8%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PSIP1	100%	100%	100%	100%	99.9%	
PSKH1	100%	100%	100%	100%	99.4%	Cholestasis, progressive familial intrahepatic, 13, 620962
PSMA3	95.6%	95.6%	100%	100%	99.8%	
PSMB1	90.4%	90.4%	100%	100%	99.8%	?Neurodevelopmental disorder with microcephaly, hypotonia, and absent language, 620038
PSMB10	100%	100%	100%	100%	99%	Immunodeficiency 121 with autoinflammation, 620807;Proteasome-associated autoinflammatory syndrome 5, 619175
PSMB4	100%	100%	100%	100%	99.8%	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms, 617591
PSMB8	100%	100%	100%	99.9%	99.2%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSMB9	100%	100%	100%	100%	99.7%	Proteasome-associated autoinflammatory syndrome 6, 620796
PSMC1	100%	100%	100%	99.9%	99.6%	?Birk-Aharoni syndrome, 620071
PSMC3	100%	100%	100%	99.9%	99.1%	Ebstein-Bezieau neurodevelopmental syndrome, 621539;?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354
PSMC3IP	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 3, 614324
PSMC5	100%	100%	100%	100%	99.2%	Yu-Kury neurodevelopmental syndrome, 621565

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PSMD11	94.1%	94.1%	100%	100%	99.8%	
PSMD12	89.3%	89.3%	100%	100%	99.8%	Stankiewicz-Isidor syndrome, 617516
PSMG2	70.6%	70.6%	100%	100%	99.6%	?Proteasome-associated autoinflammatory syndrome 4, 619183
PSPH	100%	100%	100%	100%	99.8%	Phosphoserine phosphatase deficiency, 614023
PSTPIP1	100%	100%	100%	100%	99.7%	Autoinflammatory syndrome with cytopenia, hyperzincemia, and hypercalprotectinemia, 601979;Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTBP1	100%	99.3%	100%	100%	99.4%	STAD syndrome, 621495
PTCD3	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 51, 619057
PTCH1	100%	100%	100%	100%	99.2%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTCH2	100%	100%	100%	99.9%	99.4%	Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462
PTCHD1	100%	100%	98.9%	89.1%	69.9%	{Autism, susceptibility to, X-linked 4}, 300830
PTCRA	100%	100%	100%	100%	99.8%	Immunodeficiency 126, 620931
PTDSS1	93%	93%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PTEN	89.6%	89.6%	100%	100%	99.6%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTF1A	100%	100%	100%	99.5%	97.6%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935
PTGIS	100%	100%	100%	100%	99.6%	Hypertension, essential, 145500
PTGS1	96.6%	96.6%	100%	100%	99.7%	
PTH	100%	100%	100%	100%	100%	Hypoparathyroidism, familial isolated 1, 146200
PTH1R	100%	100%	100%	100%	99%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
PTHLH	100%	100%	100%	100%	99.8%	Brachydactyly, type E2, 613382
PTPA	100%	100%	100%	100%	99.1%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482
PTPMT1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with ataxia and brain abnormalities, 621199

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN12	100%	100%	100%	100%	99.6%	Colon cancer, somatic, 114500
PTPN14	100%	100%	100%	100%	99.5%	Choanal atresia and lymphedema, 613611
PTPN22	96.8%	96.8%	100%	100%	99.9%	
PTPN23	100%	100%	100%	100%	99%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPN4	100%	100%	100%	100%	99.9%	
PTPRC	100%	100%	100%	100%	99.8%	Immunodeficiency 105, severe combined, 619924
PTPRF	100%	100%	100%	100%	99.4%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTPRJ	100%	100%	100%	99.9%	99.5%	Colon cancer, somatic, 114500;Thrombocytopenia 10, 620484
PTPRO	100%	100%	100%	100%	99.8%	Nephrotic syndrome, type 6, 614196
PTPRQ	100%	100%	100%	99.9%	99.7%	Deafness, autosomal dominant 73, 617663;Deafness, autosomal recessive 84A, 613391
PTRH2	100%	100%	100%	100%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PTRHD1	100%	100%	100%	99.8%	99.4%	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747
PTS	100%	100%	100%	100%	99.6%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100%	100%	100%	100%	99.2%	Verheij syndrome, 615583
PUM1	92.4%	92.4%	100%	99.9%	99.2%	Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719
PURA	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100%	100%	100%	99.9%	99.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	100%	100%	100%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PXDN	100%	99.6%	100%	99.9%	99.3%	Anterior segment dysgenesis 7, with sclerocornea, 269400
PYCR1	100%	100%	100%	100%	99.8%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420
PYGL	100%	100%	100%	99.9%	99.4%	Glycogen storage disease VI, 232700
PYGM	100%	100%	100%	100%	99.6%	McArdle disease, 232600
PYROXD1	92.6%	92.6%	100%	100%	99.6%	Myopathy, myofibrillar, 8, 617258
PYROXD2	82.2%	79.5%	100%	99.9%	99.1%	
QARS1	100%	100%	100%	100%	99.2%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	93.7%	93.6%	100%	100%	99.3%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100%	100%	100%	100%	99.4%	Ververi-Brady syndrome 1, 617982
QRICH2	100%	100%	100%	100%	99.6%	Spermatogenic failure 35, 618341
QRSL1	91.9%	89.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 40, 618835
RAB11A	92.9%	92.9%	100%	100%	99.6%	
RAB11B	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100%	100%	100%	100%	99.6%	
RAB18	100%	100%	100%	100%	99.5%	Warburg micro syndrome 3, 614222
RAB23	100%	100%	100%	100%	99.4%	Carpenter syndrome 1, 201000
RAB27A	100%	100%	100%	100%	99.6%	Griscelli syndrome, type 2, 607624
RAB28	100%	100%	100%	100%	99.6%	Cone-rod dystrophy 18, 615374

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RAB32	100%	100%	100%	99.9%	99.3%	{Parkinson disease 26, autosomal dominant, susceptibility to}, 620923
RAB33B	100%	100%	100%	100%	99.4%	Smith-McCort dysplasia 2, 615222
RAB34	100%	100%	100%	100%	99.5%	Orofaciodigital syndrome XX, 620718
RAB39B	100%	100%	99.4%	90.8%	72.7%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RAB3GAP1	95.7%	95.7%	100%	100%	99.6%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	94.4%	94.4%	100%	100%	99.8%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RAB5C	100%	100%	100%	99.9%	99.2%	
RAB5IF	100%	100%	100%	99.8%	99.3%	?Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development syndrome 2, 616994
RAB7A	89.2%	89.2%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 2B, 600882
RABGAP1	90.8%	90.2%	100%	100%	99.8%	
RAC1	78.8%	78.8%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 48, 617751

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RAC2	100%	100%	100%	99.9%	99.4%	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203;?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986
RAC3	98.6%	91.2%	100%	100%	99.2%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RACGAP1	95.4%	92.8%	100%	100%	99.7%	Anemia, congenital dyserythropoietic, type IIIb, autosomal recessive, 619789
RAD21	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD21L1	100%	100%	100%	100%	99.8%	
RAD50	100%	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078
RAD51	89.4%	89.4%	100%	100%	99.5%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAD51C	87.3%	85.8%	100%	100%	99.2%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RAD51D	100%	100%	100%	100%	99.8%	{Breast-ovarian cancer, familial, susceptibility to, 4}, 614291
RAD54B	96.5%	96.5%	100%	100%	99.7%	Colon cancer, somatic, 114500;Lymphoma, non-Hodgkin, somatic, 605027
RAD54L	100%	100%	100%	100%	99.6%	{Breast cancer, invasive ductal}, 114480;Adenocarcinoma, colonic, somatic;Lymphoma, non-Hodgkin, somatic, 605027
RAF1	96.1%	93.8%	100%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RAG1	100%	100%	100%	100%	99.7%	Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	100%	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RAI1	100%	100%	100%	100%	99%	Smith-Magenis syndrome, 182290

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RALA	100%	100%	100%	99.9%	99.8%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RANBP2	100%	100%	100%	100%	99.8%	{Encephalopathy, acute, infection-induced, 3, susceptibility to}, 608033
RANGRF	100%	100%	100%	99.9%	99.2%	
RAP1B	100%	100%	100%	100%	99.6%	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654
RAP1GDS1	94.7%	94.7%	100%	100%	99.9%	Alfadhel syndrome, 620655
RAPGEF2	100%	100%	100%	100%	99.6%	?Epilepsy, familial adult myoclonic, 7, 618075
RAPSN	100%	100%	100%	100%	99.6%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RARB	98%	98%	100%	100%	99.7%	Microphthalmia, syndromic 12, 615524
RARS1	90.9%	90.5%	100%	99.9%	99.4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	94.2%	93.8%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 6, 611523

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RASA1	100%	100%	100%	100%	99.8%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RASA2	96.6%	96.6%	100%	100%	99.7%	
RASGRP1	95%	95%	100%	100%	99.8%	Immunodeficiency 64, 618534
RASGRP2	98.6%	98%	100%	100%	99.4%	Bleeding disorder, platelet-type, 18, 615888
RAX	100%	100%	100%	100%	99.1%	Microphthalmia, syndromic 16, 611038
RAX2	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 95, 620102;Cone-rod dystrophy 11, 610381;?Macular degeneration, age-related, 6, 613757
RB1	96%	96%	100%	100%	99.8%	Small cell cancer of the lung, somatic, 182280;Bladder cancer, somatic, 109800;Retinoblastoma, trilateral, 180200;Osteosarcoma, somatic, 259500;Retinoblastoma, 180200
RB1CC1	100%	100%	100%	100%	99.8%	Breast cancer, somatic, 114480
RBBP5	100%	100%	100%	100%	99.5%	
RBBP6	100%	100%	100%	100%	99.6%	
RBBP7	95.1%	94.2%	99.3%	90.3%	71.8%	Spermatogenic failure, X-linked, 9, 301137
RBBP8	100%	100%	100%	100%	99.8%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RBCK1	100%	100%	100%	99.8%	98.8%	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895
RBFOX1	100%	99.7%	100%	99.9%	99.3%	
RBFOX2	85.5%	84.7%	100%	100%	99.6%	
RBL2	100%	100%	100%	100%	99.6%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM10	95.6%	95.6%	98.7%	88.7%	70.4%	TARP syndrome, 311900
RBM20	100%	100%	100%	100%	99.4%	Cardiomyopathy, dilated, 1DD, 613172
RBM28	100%	100%	100%	100%	99.7%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBM8A	100%	100%	100%	99.7%	98.9%	Thrombocytopenia-absent radius syndrome, 274000
RBMX	100%	100%	99.4%	93.8%	78.6%	?Intellectual developmental disorder, X-linked syndromic, Gustavson type, 309555;?Intellectual developmental disorder, X-linked syndromic, Shashi type, 300238
RBMY1A1	50%	50%	50%	49.2%	46.1%	
RBMY1B	50%	50%	49%	45.6%	40.4%	
RBMY1D	49.8%	48.7%	47%	44.5%	39.3%	
RBMY1E	50%	50%	49.2%	47.3%	42.5%	
RBMY1F	49.7%	49.6%	48.3%	43.5%	31.3%	
RBMY1J	50%	50%	49.2%	44.7%	32.3%	
RBP3	100%	100%	100%	100%	99.5%	?Retinitis pigmentosa 66, 615233

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RBP4	100%	100%	100%	100%	98.8%	Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	100%	100%	100%	100%	99.6%	Adams-Oliver syndrome 3, 614814
RBSN	96.1%	96.1%	100%	100%	99.5%	Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad neurodevelopmental syndrome, 620937
RC3H1	100%	100%	100%	100%	99.7%	?Immune dysregulation and systemic hyperinflammation syndrome, 618998
RCBTB1	100%	100%	100%	100%	99.7%	Retinal dystrophy with or without extraocular anomalies, 617175
RCC1	100%	100%	100%	99.9%	99.3%	Infection-induced acute-onset axonal neuropathy, 621333
RD3	100%	100%	100%	100%	99.4%	Leber congenital amaurosis 12, 610612
RDH11	94.9%	94%	100%	100%	99.5%	?Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108
RDH12	100%	100%	100%	100%	99.6%	Leber congenital amaurosis 13, 612712
RDH5	100%	100%	100%	100%	99.5%	Fundus albipunctatus, 136880
RDH8	100%	100%	100%	100%	99%	Stargardt disease 5, 621259
RDX	100%	100%	100%	100%	99.9%	Deafness, autosomal recessive 24, 611022

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

REC114	100%	100%	100%	100%	100%	Oocyte/zygote/embryo maturation arrest 10, 619176
RECQL	97.9%	97.9%	100%	100%	99.7%	RECON progeroid syndrome, 620370
RECQL4	100%	100%	100%	100%	99.4%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
REEP1	87.5%	87.5%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751
REEP2	100%	100%	100%	99.8%	99.3%	Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606
REEP6	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 77, 617304
REL	100%	100%	100%	100%	99.7%	Immunodeficiency 92, 619652
RELA	100%	100%	100%	100%	99.6%	Autoinflammatory disease, familial, Behcet-like-3, 618287
RELB	100%	100%	100%	99.9%	98.6%	Immunodeficiency 53, 617585
RELN	100%	100%	100%	100%	99.8%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320
RELT	100%	100%	100%	100%	99.7%	Amelogenesis imperfecta, type IIIC, 618386

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

REN	100%	100%	100%	100%	99.4%	Renal tubular dysgenesis, 267430;[Hyperprorenin emia];Tubulointerstitial kidney disease, autosomal dominant, 4, 613092
REPS1	100%	100%	100%	100%	99.9%	?Neurodegeneration with brain iron accumulation 7, 617916
RERE	99%	99%	100%	99.8%	98.2%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REST	100%	100%	100%	99.9%	99.5%	Deafness, autosomal dominant 27, 612431;{Wilms tumor 6, susceptibility to}, 616806;Fibromatosis, gingival, 5, 617626
RET	100%	100%	100%	100%	99.4%	{Hirschsprung disease, susceptibility to, 1}, 142623;Multiple endocrine neoplasia IIA, 171400;{Hirschsprung disease, protection against}, 142623;Medullary thyroid carcinoma, 155240;Pheochromocyt oma, 171300;Multiple endocrine neoplasia IIB, 162300
RETREG1	91.3%	91.3%	100%	99.9%	99.5%	Neuropathy, hereditary sensory and autonomic, type IIB, 613115
REV3L	100%	100%	100%	100%	99.8%	
RFC1	100%	100%	100%	100%	99.7%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RFC4	100%	100%	100%	100%	99.8%	Morimoto-Ryu-Malicda n neuromuscular syndrome, 621010

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RFT1	84.9%	84.9%	100%	100%	99.8%	Congenital disorder of glycosylation, type In, 612015
RFWD3	100%	100%	100%	100%	99.8%	?Fanconi anemia, complementation group W, 617784
RFX3	100%	100%	100%	100%	99.9%	
RFX4	100%	100%	100%	99.9%	99.1%	
RFX5	100%	100%	100%	100%	99.8%	?MHC class II deficiency 5, 620818;MHC class II deficiency 3, 620816
RFX6	100%	100%	100%	100%	99.8%	Mitchell-Riley syndrome, 615710
RFX7	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330
RFXANK	100%	100%	100%	100%	99.4%	MHC class II deficiency 2, 620815
RFXAP	100%	100%	100%	99.8%	98.8%	MHC class II deficiency 4, 620817
RGR	100%	100%	100%	100%	99.1%	Retinitis pigmentosa 44, 613769
RGS10	100%	100%	100%	99.9%	99.4%	
RGS12	100%	100%	100%	100%	99.5%	
RGS9	95.9%	95.9%	100%	100%	99.6%	Prolonged electroretinal response suppression 1, 608415
RGS9BP	100%	100%	100%	100%	99.4%	Prolonged electroretinal response suppression 2, 620344
RHAG	100%	100%	100%	100%	99.5%	Overhydrated hereditary stomatocytosis, 185000;Anemia, hemolytic, Rh-null, regulator type, 268150

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RHBDF2	100%	100%	100%	99.9%	98.5%	Tylosis with esophageal cancer, 148500
RHCE	97.1%	97.1%	96.8%	95.5%	91.9%	Rh-null disease, amorph type, 617970
RHEB	100%	100%	100%	100%	99.7%	
RHO	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary, autosomal dominant 1, 610445;Retinitis pigmentosa 4, autosomal dominant or recessive, 613731;Retinitis punctata albescens, 136880
RHOA	81.7%	81.7%	100%	100%	99.6%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RHOBTB2	98.7%	98.7%	100%	100%	99.6%	Developmental and epileptic encephalopathy 64, 618004
RHOG	100%	100%	100%	100%	100%	
RHOH	100%	100%	100%	100%	99.1%	Immunodeficiency 129, 618307
RIC1	100%	100%	100%	100%	99.8%	CATIFA syndrome, 618761
RICTOR	100%	100%	100%	100%	99.7%	
RIGI	98.6%	98.6%	100%	100%	99.9%	Singleton-Merten syndrome 2, 616298
RILPL1	87.8%	87.8%	100%	100%	99.4%	Oculopharyngodistal myopathy 4, 619790
RIMS1	100%	100%	100%	100%	99.6%	
RIMS2	99.2%	99.2%	100%	100%	99.8%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RIN2	98.3%	98.3%	100%	100%	99.2%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RINT1	93.6%	93.6%	100%	100%	99.8%	Infantile liver failure syndrome 3, 618641
RIPK1	100%	99.8%	100%	100%	99.5%	Immunodeficiency 57 with autoinflammation, 618108;Autoinflammation with episodic fever and lymphadenopathy, 618852
RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIPOR2	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 21, 607017;?Deafness, autosomal recessive 104, 616515
RIPPLY2	100%	100%	100%	100%	99.9%	Spondylocostal dysostosis 6, 616566
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355
RLBP1	100%	100%	100%	100%	99.8%	Bothnia retinal dystrophy, 607475;Newfoundland rod-cone dystrophy, 607476;Retinitis punctata albescens, 136880;Fundus albipunctatus, 136880
RLF	100%	100%	100%	100%	99.9%	
RLIM	100%	100%	98.9%	91.9%	75%	Tonne-Kalscheuer syndrome, 300978
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNASEH1	88.3%	88.3%	100%	100%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479
RNASEH2A	100%	100%	100%	100%	98.8%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.5%	91.5%	100%	99.9%	99.5%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	99.9%	98.6%	Aicardi-Goutieres syndrome 3, 610329
RNASEL	100%	100%	100%	100%	99.6%	Prostate cancer 1, 601518
RNASET2	79.4%	74.3%	100%	100%	99.4%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF111	97.5%	97.5%	100%	100%	99.6%	
RNF113A	100%	100%	99.3%	87.4%	70.4%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100%	100%	100%	100%	99.5%	Tenorio syndrome, 616260
RNF13	85.4%	85.3%	100%	100%	99.8%	Developmental and epileptic encephalopathy 73, 618379
RNF135	96%	96%	100%	100%	99.9%	
RNF139	100%	100%	100%	100%	99.7%	Renal cell carcinoma, 144700
RNF168	100%	100%	100%	100%	99.3%	RIDDLE syndrome, 611943

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RNF170	100%	100%	100%	100%	99.9%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686
RNF2	100%	100%	100%	100%	99.5%	Luo-Schoch-Yamamoto syndrome, 619460
RNF212	100%	100%	100%	99.9%	99%	?Spermatogenic failure 62, 619673;Recombination rate QTL 1, 612042
RNF212B	100%	100%	100%	99.9%	99.6%	
RNF213	100%	100%	100%	100%	99.6%	{Moyamoya disease 2, susceptibility to}, 607151
RNF216	100%	100%	100%	100%	99.7%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RNF220	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNF31	100%	99.9%	100%	100%	99.2%	Immunodeficiency 115 with autoinflammation, 620632
RNF43	100%	100%	100%	100%	99.5%	Sessile serrated polyposis cancer syndrome, 617108
RNF6	100%	100%	100%	100%	99.8%	Esophageal carcinoma, somatic, 133239
RNPC3	100%	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU12						CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RNU2-2						Developmental and epileptic encephalopathy 119, 621304
RNU4-2						Retinitis pigmentosa 102, 621560;ReNU syndrome, 620851
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU5B-1						Neurodevelopmental disorder with seizures and joint laxity, 621302
RNU6-1						Retinitis pigmentosa 103, 621561
RNU6-2						Retinitis pigmentosa 104, 621562
RNU6-8						Retinitis pigmentosa 105, 621563
RNU6-9						Retinitis pigmentosa 106, 621564
RNU6ATAC						
RNU7-1						Aicardi-Goutieres syndrome 9, 619487
ROBO1	100%	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROBO2	100%	100%	100%	100%	99.8%	Vesicoureteral reflux 2, 610878
ROBO3	100%	100%	100%	100%	99.4%	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ROBO4	100%	100%	100%	100%	99.4%	Aortic valve disease 3, 618496
ROGDI	100%	100%	100%	99.9%	98.8%	Kohlschutter-Tonz syndrome, 226750
ROM1	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 7, digenic form, 608133
ROR1	100%	100%	100%	100%	99.8%	?Deafness, autosomal recessive 108, 617654
ROR2	100%	100%	100%	100%	99.6%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310
RORA	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORB	100%	100%	100%	100%	99.9%	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357
RORC	100%	100%	100%	100%	99.5%	Immunodeficiency 42, 616622
RP1	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 1, 180100
RP1L1	100%	100%	100%	99.9%	99%	Occult macular dystrophy, 613587;Retinitis pigmentosa 88, 618826
RP2	100%	100%	98.2%	89.2%	73.9%	Retinitis pigmentosa 2, 312600
RP9	100%	100%	100%	99.8%	98%	?Retinitis pigmentosa 9, 180104
RPA1	93.6%	93.3%	100%	100%	99.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RPE65	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 20, 613794;Retinitis pigmentosa 87 with choroidal involvement, 618697;Leber congenital amaurosis 2, 204100
RPGR	100%	99.7%	92.5%	79.2%	61.2%	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455;Cone-rod dystrophy, X-linked, 1, 304020;Retinitis pigmentosa 3, 300029;Macular degeneration, X-linked atrophic, 300834
RPGRIP1	100%	100%	100%	99.9%	99.1%	Cone-rod dystrophy 13, 608194;Leber congenital amaurosis 6, 613826
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPH3A	100%	100%	100%	99.9%	99.7%	
RPIA	100%	100%	100%	100%	99.7%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100%	100%	99.3%	91.1%	69.5%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPL10A	100%	100%	100%	100%	99.2%	
RPL10L	100%	100%	100%	100%	99.7%	?Spermatogenic failure 63, 619689
RPL11	96.3%	94.4%	100%	100%	99.9%	Diamond-Blackfan anemia 7, 612562

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RPL13	100%	100%	100%	100%	98.9%	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type, 618728
RPL15	100%	98.5%	100%	100%	100%	Diamond-Blackfan anemia 12, 615550
RPL17	100%	100%	100%	100%	99.7%	Diamond-Blackfan anemia 22, 621262
RPL18	100%	100%	100%	100%	99.6%	?Diamond-Blackfan anemia 18, 618310
RPL21	100%	100%	100%	100%	99.8%	Hypotrichosis 12, 615885
RPL26	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 11, 614900
RPL27	100%	100%	100%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL31	91.5%	84.2%	100%	99.8%	99.4%	
RPL35	82.1%	82.1%	100%	100%	99.7%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	100%	100%	99.1%	Diamond-Blackfan anemia 5, 612528
RPL3L	100%	99.9%	100%	99.8%	99.2%	Cardiomyopathy, dilated, 2D, 619371
RPL4	100%	100%	100%	100%	99.8%	
RPL5	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	100%	100%	99.9%	
RPN2	96.5%	96.5%	100%	100%	99.8%	
RPS10	89.1%	89.1%	100%	100%	99.7%	Diamond-Blackfan anemia 9, 613308
RPS14	82.1%	75.4%	100%	99.9%	99.4%	Macrocytic anemia, refractory, due to 5q deletion, somatic, 153550
RPS15A	63%	63%	100%	100%	99.6%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 4, 612527

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RPS19	100%	100%	100%	100%	99.2%	Diamond-Blackfan anemia 1, 105650
RPS20	100%	100%	100%	100%	99.6%	
RPS23	100%	100%	100%	100%	99.8%	Brachycephaly, trichomegaly, and developmental delay, 617412
RPS24	88.4%	86.1%	100%	100%	99.7%	Diamond-blackfan anemia 3, 610629
RPS26	100%	99.9%	100%	100%	99.7%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	100%	100%	99.7%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	100%	100%	99.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	100%	100%	99.4%	Diamond-Blackfan anemia 13, 615909
RPS4Y2	50%	50%	48.4%	38.8%	21.6%	
RPS6KA3	97.6%	97.6%	99.2%	92.4%	75.4%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600
RPS6KB1	100%	100%	100%	100%	99.5%	
RPS6KC1	97.6%	97.6%	100%	100%	99.5%	Neurodevelopmental disorder with spasticity, thin corpus callosum, and decreased brain white matter, 621460
RPS7	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 8, 612563
RPSA	100%	100%	100%	100%	99.9%	Asplenia, isolated congenital, 271400
RRAD	100%	100%	100%	100%	99.4%	
RRAGC	100%	100%	100%	100%	99.5%	Long-Olsen-Distelmaier syndrome, 620609

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RRAGD	100%	100%	100%	100%	99.8%	Hypomagnesemia 7, renal, with or without dilated cardiomyopathy, 620152
RRAS	93.2%	93.2%	100%	99.9%	98.4%	
RRAS2	95.5%	95.5%	100%	100%	99.5%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	99.9%	98.9%	
RRM1	100%	100%	100%	100%	99.7%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 6, 620647
RRM2B	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP12	100%	100%	100%	100%	99.2%	Basal ganglia calcification, idiopathic, 11, autosomal recessive, 621452
RRP7A	100%	100%	99.9%	99.3%	96.9%	?Microcephaly 28, primary, autosomal recessive, 619453
RS1	100%	100%	98.6%	87.3%	69.1%	Retinoschisis, 312700
RSF1	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RSPH1	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 12, 612650
RSPO1	100%	100%	100%	100%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO2	100%	100%	100%	99.9%	99.5%	?Humerofemoral hypoplasia with radiotibial ray deficiency, 618022;Tetraamelia syndrome 2, 618021
RSPO4	100%	100%	100%	100%	99.4%	Anonychia congenita, 206800
RSPRY1	95.5%	95.5%	100%	100%	99.4%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	84.2%	84.2%	100%	99.9%	99.5%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100%	100%	100%	99.9%	99.4%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RTN2	100%	100%	100%	100%	99.2%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805
RTN4IP1	100%	100%	100%	100%	99.8%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RTTN	100%	100%	100%	100%	99.8%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100%	100%	100%	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1	100%	100%	100%	99.7%	97.9%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
RUNX1T1	100%	100%	100%	100%	99.7%	
RUNX2	100%	100%	100%	99.9%	98.8%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, 119600
RUSC2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 61, 617773
RXFP2	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

RXYLT1	100%	100%	100%	99.8%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYBP	100%	100%	100%	99.9%	99.4%	
RYR1	100%	100%	100%	99.9%	99%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600
RYR2	99.8%	99.8%	100%	100%	99.8%	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772;Ventricular arrhythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, 115000
RYR3	100%	100%	100%	100%	99.7%	Congenital myopathy 20, 620310
S100A3	100%	100%	100%	100%	99.4%	
S1PR2	100%	100%	100%	100%	99.7%	Deafness, autosomal recessive 68, 610419
SACS	99%	99%	100%	100%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAG	100%	100%	100%	100%	99.6%	Retinitis pigmentosa 47, autosomal recessive, 613758;Retinitis pigmentosa 96, autosomal dominant, 620228;Oguchi disease-1, 258100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SALL1	100%	100%	100%	100%	99.2%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SALL2	100%	100%	100%	100%	99.7%	?Coloboma, ocular, autosomal recessive, 216820
SALL4	100%	100%	100%	100%	99.6%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323
SAMD11	100%	100%	100%	100%	98.9%	
SAMD12	100%	100%	100%	100%	99.7%	Epilepsy, familial adult myoclonic, 1, 601068
SAMD7	100%	100%	100%	100%	99.2%	Macular dystrophy with or without cone dysfunction, 620762
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	92%	89.8%	100%	100%	99.5%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SAR1B	88.5%	84.5%	100%	100%	98.9%	Chylomicron retention disease, 246700
SARDH	84.7%	84.7%	100%	100%	99.5%	[Sarcosinemia], 268900
SARS1	89.1%	89.1%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SARS2	100%	100%	100%	100%	99.7%	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845
SART3	100%	99.6%	100%	100%	99.4%	
SASH1	98.3%	97.2%	100%	100%	99.5%	Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SASH3	100%	100%	99.1%	89.2%	70.5%	Immunodeficiency 102, 301082
SASS6	100%	100%	100%	100%	99.7%	Microcephaly 14, primary, autosomal recessive, 616402
SAT1	100%	100%	98.9%	89.4%	69.8%	
SATB1	100%	100%	100%	100%	99.7%	den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100%	100%	100%	99.9%	99.5%	Glass syndrome, 612313
SBDS	100%	100%	100%	100%	99.9%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SBF1	100%	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, type 4B3, 615284
SBF2	93.7%	93.7%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 4B2, 604563
SC5D	100%	100%	100%	100%	99.9%	Lathosterolosis, 607330
SCAF4	100%	100%	100%	100%	99.3%	Fliedner-Zweier syndrome, 620511
SCAMP5	100%	100%	100%	100%	99.5%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SCAPER	97.9%	97.9%	100%	100%	99.8%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCARB2	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCARF2	100%	100%	100%	99.8%	97.6%	Van den Ende-Gupta syndrome, 600920
SCD5	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 79, 619086
SCLT1	95.2%	95.2%	100%	100%	99.8%	
SCN10A	100%	100%	100%	100%	99.8%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100%	100%	100%	99.8%	99.4%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317;Migraine, familial hemiplegic, 3, 609634;Dravet syndrome, 607208;Febrile seizures, familial, 3A, 604403;Generalized epilepsy with febrile seizures plus, type 2, 604403

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SCN1B	100%	100%	100%	100%	99.5%	Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838
SCN2A	100%	100%	100%	100%	99.8%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
SCN2B	100%	100%	100%	99.9%	99%	Atrial fibrillation, familial, 14, 615378
SCN3A	100%	100%	100%	100%	99.8%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN3B	90%	89.7%	100%	100%	99.7%	Atrial fibrillation, familial, 16, 613120;Brugada syndrome 7, 613120
SCN4A	100%	100%	100%	100%	99.4%	Paramyotonia congenita, 168300;Hyperkalemic periodic paralysis, 170500;Congenital myopathy 22B, severe fetal, 620369;Hypokalemic periodic paralysis, type 2, 613345;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Myasthenic syndrome, congenital, 16, 614198;Congenital myopathy 22A, classic, 620351

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SCN4B	100%	100%	100%	100%	99.2%	Atrial fibrillation, familial, 17, 611819;Long QT syndrome 10, 611819
SCN5A	100%	100%	100%	100%	99.5%	Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120
SCN7A	100%	100%	100%	99.9%	99.8%	
SCN8A	100%	100%	100%	100%	99.6%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558
SCN9A	100%	100%	100%	100%	99.5%	Erythermalgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000
SCNM1	100%	100%	100%	99.7%	98.4%	Orofaciodigital syndrome XIX, 620107

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SCNN1A	100%	100%	100%	99.9%	99.5%	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350;?Liddle syndrome 3, 618126;Bronchiectasis with or without elevated sweat chloride 2, 613021
SCNN1B	100%	100%	100%	100%	99.6%	Bronchiectasis with or without elevated sweat chloride 1, 211400;Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125;Liddle syndrome 1, 177200
SCNN1G	100%	100%	100%	100%	99.2%	Bronchiectasis with or without elevated sweat chloride 3, 613071;Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126;Liddle syndrome 2, 618114
SCO1	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	100%	100%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCP2	95.5%	95.5%	100%	100%	99.7%	?Leukoencephalopathy with dystonia and motor neuropathy, 613724
SCUBE3	97.5%	97.5%	100%	100%	99.8%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 2, 619184
SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SCYL2	97.3%	97.3%	100%	100%	99.9%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	100%	100%	100%	100%	99.9%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SDHA	92.6%	89.3%	100%	100%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165
SDHAF1	100%	100%	100%	100%	99.3%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SDHAF2	100%	99.7%	100%	100%	99.6%	Pheochromocytoma/paraganglioma syndrome 2, 601650
SDHB	94%	94%	100%	100%	99.7%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHC	77.5%	77.5%	100%	100%	99.9%	Pheochromocytoma/paraganglioma syndrome 3, 605373;Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SDHD	78.9%	78.9%	100%	100%	99.5%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
SDR9C7	100%	100%	100%	100%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23A	100%	100%	100%	100%	99.7%	Craniolenticulosutural dysplasia, 607812
SEC23B	100%	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SEC24D	100%	100%	100%	100%	99.6%	Cole-Carpenter syndrome 2, 616294
SEC31A	96.6%	96.6%	100%	99.9%	99.7%	?Halperin-Birk syndrome, 618651
SEC61A1	92.3%	92.3%	100%	99.9%	99.4%	Immunodeficiency, common variable, 15, 620670;?Neutropenia, severe congenital, 11, autosomal dominant, 620674;Tubulointerstitial kidney disease, autosomal dominant, 5, 617056
SEC61B	100%	100%	100%	100%	99%	
SEC63	92.9%	92.9%	100%	100%	99.9%	Polycystic liver disease 2, 617004
SECISBP2	100%	100%	100%	100%	99.7%	Thyroid hormone metabolism, abnormal, 1, 609698

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SEL1L	96.7%	96.7%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, poor growth, dysmorphic facies, and agammaglobulinemia, 621068;?Neurodevelopmental disorder with poor growth, absent speech, progressive ataxia, and dysmorphic facies, 621067
SELENBP1	93.1%	93.1%	100%	100%	99.4%	Extraoral halitosis due to MTO deficiency, 618148
SELENOI	100%	100%	100%	100%	99.9%	Spastic paraplegia 81, autosomal recessive, 618768
SELENON	93.9%	91.8%	100%	99.8%	99.3%	Congenital myopathy 3 with rigid spine, 602771
SEMA3A	100%	100%	100%	100%	99.8%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100%	100%	100%	100%	99.9%	
SEMA3F	100%	100%	100%	99.9%	99.1%	
SEMA4A	100%	100%	100%	100%	98.9%	Retinitis pigmentosa 35, 610282;Cone-rod dystrophy 10, 610283
SEMA6A	100%	100%	100%	100%	99.6%	
SEMA6B	100%	100%	100%	99.8%	98.9%	Epilepsy, progressive myoclonic, 11, 618876
SEMA7A	100%	100%	100%	100%	99.5%	?Cholestasis, progressive familial intrahepatic, 11, 619874;[Blood group, John-Milton-Hagen system], 614745
SEPHS1	91.3%	86.6%	100%	100%	99.8%	Ververi-Brady syndrome 2, 621325
SEPSECS	99.4%	96.3%	100%	100%	99.5%	Pontocerebellar hypoplasia type 2D, 613811

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SEPTIN12	100%	100%	100%	100%	99%	Spermatogenic failure 10, 614822
SEPTIN2	93.9%	93.9%	100%	100%	99.7%	
SEPTIN4	100%	100%	100%	99.9%	99.3%	Spermatogenic failure 99, 621194
SEPTIN9	100%	100%	100%	99.9%	98.9%	Amyotrophy, hereditary neuralgic, 162100
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SERPINA1	100%	100%	100%	100%	99.3%	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490;Emphysema due to AAT deficiency, 613490;Emphysema-cirrhosis, due to AAT deficiency, 613490
SERPINA12	100%	100%	100%	100%	99.8%	
SERPINA3	100%	100%	100%	100%	99.9%	Alpha-1-antichymotrypsin deficiency;Cerebrovascular disease, occlusive
SERPINA6	93%	93%	100%	100%	99.8%	Corticosteroid-binding globulin deficiency, 611489
SERPINB6	91%	90.7%	100%	100%	99.5%	?Deafness, autosomal recessive 91, 613453
SERPINB7	100%	100%	100%	100%	99.7%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100%	100%	100%	100%	99.6%	Peeling skin syndrome 5, 617115
SERPINC1	90.4%	88.6%	100%	100%	99.9%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPIND1	100%	100%	100%	100%	99.8%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SERPINE1	100%	100%	100%	100%	99.6%	Plasminogen activator inhibitor-1 deficiency, 613329;{Transcription of plasminogen activator inhibitor, modulator of}
SERPINF1	100%	100%	100%	100%	99%	Osteogenesis imperfecta, type VI, 613982
SERPINF2	92.1%	91.2%	100%	100%	99.4%	Alpha-2-plasmin inhibitor deficiency, 262850
SERPING1	100%	100%	100%	100%	99.6%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790
SERPINH1	100%	100%	100%	100%	99.6%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SERPINI1	100%	100%	100%	100%	99.9%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SET	100%	100%	100%	99.9%	99.2%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	100%	100%	100%	100%	99.5%	Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078
SETD1A	100%	100%	100%	100%	99.1%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SETD1B	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100%	100%	100%	100%	99.7%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SETD5	99.7%	99.1%	100%	99.9%	99.5%	Intellectual developmental disorder, autosomal dominant 23, 615761
SETX	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SEZ6	100%	100%	100%	99.9%	99.3%	
SF1	100%	100%	100%	99.7%	98.7%	
SF3B1	100%	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286
SF3B2	100%	100%	100%	100%	99.4%	Craniofacial microsomia, 164210
SF3B3	98%	96.9%	100%	100%	99.6%	
SF3B4	100%	100%	100%	100%	99.1%	Acrofacial dysostosis 1, Nager type, 154400
SFRP4	100%	100%	100%	100%	99.8%	Pyle disease, 265900
SFTPA1	96.2%	95.1%	100%	100%	99.8%	Interstitial lung disease 1, 619611
SFTPA2	100%	100%	100%	100%	99.5%	Interstitial lung disease 2, 178500
SFTPB	100%	100%	100%	100%	99.3%	Surfactant metabolism dysfunction, pulmonary, 1, 265120

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SFTPC	100%	100%	100%	100%	99.5%	Surfactant metabolism dysfunction, pulmonary, 2, 610913
SFXN4	100%	100%	100%	99.9%	99.6%	Combined oxidative phosphorylation deficiency 18, 615578
SGCA	97.5%	97.5%	100%	100%	99.2%	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099
SGCB	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286
SGCD	94%	94%	100%	100%	99.7%	Cardiomyopathy, dilated, 1L, 606685;Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287
SGCE	89.5%	89.5%	100%	100%	99.6%	Dystonia-11, myoclonic, 159900
SGCG	83.2%	83.2%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700
SGMS1	100%	100%	100%	100%	99.5%	
SGMS2	100%	100%	100%	100%	99.4%	Calvarial doughnut lesions with bone fragility with or without spondylometaphyseal dysplasia, 126550
SGO1	100%	100%	100%	100%	99.9%	Chronic atrial and intestinal dysrhythmia, 616201
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SGSH	100%	100%	100%	100%	99.5%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SGSM3	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 84, 620401

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SH2B3	100%	100%	100%	99.9%	99.3%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	99.4%	91.9%	73.1%	Lymphoproliferative syndrome, X-linked, 1, 308240
SH3BP2	100%	99.7%	100%	100%	99.5%	Cherubism, 118400
SH3KBP1	98.8%	98.8%	99.1%	90.5%	72.4%	?Immunodeficiency 61, 300310
SH3PXD2B	99.9%	98.6%	100%	100%	99.2%	Frank-ter Haar syndrome, 249420
SH3TC2	100%	100%	100%	100%	99.5%	Charcot-Marie-Tooth disease, type 4C, 601596;Mononeuropathy of the median nerve, mild, 613353
SHANK1	99.4%	98.7%	100%	99.8%	97.6%	
SHANK2	100%	100%	100%	100%	99%	{Autism susceptibility 17}, 613436
SHANK3	99.8%	99%	99.9%	99.7%	98.4%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SHARPIN	100%	100%	100%	100%	99.3%	Autoinflammation with episodic fever and immune dysregulation, 620795
SHH	100%	100%	100%	99.8%	98.4%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHMT2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC1	100%	100%	100%	100%	99.9%	Spermatogenic failure 75, 619949

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SHOC2	100%	100%	100%	100%	99.9%	Noonan syndrome-like with loose anagen hair 1, 607721
SHOX	47.4%	47.4%	50%	49.9%	49.6%	Short stature, idiopathic familial, 300582;Leri-Weill dyschondrosteosis, 127300;Langer mesomelic dysplasia, 249700;Short stature, idiopathic familial, 300582;Langer mesomelic dysplasia, 249700;Leri-Weill dyschondrosteosis, 127300
SHOX2	100%	100%	99.9%	99.3%	97.2%	
SHPK	100%	100%	100%	100%	99.1%	[Sedoheptulokinase deficiency], 617213
SHQ1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SHROOM3	100%	100%	100%	100%	99.6%	
SHROOM4	100%	100%	99%	90.1%	72.3%	
SI	100%	100%	100%	99.7%	99.1%	Sucrase-isomaltase deficiency, congenital, 222900
SIAH1	100%	100%	100%	100%	99.8%	Buratti-Harel syndrome, 619314
SIGMAR1	100%	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373
SIK1	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 30, 616341

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SIK3	100%	100%	100%	100%	99.4%	?Spondyloepimetaphyseal dysplasia, Krakow type, 618162
SIL1	91.5%	91.5%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800
SIN3A	97.5%	97.5%	100%	100%	99.6%	Witteveen-Kolk syndrome, 613406
SIN3B	99.2%	97.7%	100%	99.9%	99.3%	
SIPA1L3	100%	100%	100%	100%	99.4%	?Cataract 45, 616851
SIRT5	100%	100%	100%	100%	99.6%	
SIX1	100%	100%	100%	100%	98.8%	Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX3	100%	100%	100%	99.4%	97.2%	Schizencephaly, 269160;Holoprosencephaly 2, 157170
SIX5	100%	100%	100%	100%	99.4%	Branchiootorenal syndrome 2, 610896
SIX6	100%	100%	100%	100%	99.3%	Optic disc anomalies with retinal and/or macular dystrophy, 212550
SKI	100%	100%	100%	99.8%	98.8%	Shprintzen-Goldberg syndrome, 182212
SKIC2	99.7%	98.5%	100%	100%	99.6%	Trichohepatoenteric syndrome 2, 614602
SKIC3	98.9%	98.9%	100%	100%	99.9%	Trichohepatoenteric syndrome 1, 222470
SKOR2	100%	100%	100%	99.8%	97.1%	Valence-Farazi cerebellar ataxia syndrome, 621386
SLC10A1	95.7%	95.7%	100%	100%	99.3%	Hypercholanemia, familial 2, 619256
SLC10A2	100%	100%	100%	100%	99.8%	?Bile acid malabsorption, primary, 1, 613291

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC10A7	92.8%	92.8%	100%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC11A2	100%	100%	100%	100%	99.6%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC12A1	96.3%	96.3%	100%	100%	99.8%	Bartter syndrome, type 1, 601678
SLC12A2	100%	100%	100%	100%	99.6%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC12A3	94.7%	94.7%	100%	100%	99.7%	Gitelman syndrome, 263800
SLC12A5	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100%	100%	100%	100%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC12A9	100%	100%	100%	100%	98.5%	
SLC13A1	100%	100%	100%	100%	99.8%	
SLC13A3	100%	100%	100%	100%	99.6%	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate, 618384
SLC13A5	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC16A1	100%	100%	100%	100%	99.9%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC16A12	100%	100%	100%	99.9%	99.8%	Cataract 47, juvenile, with microcornea, 612018
SLC16A2	88.4%	88.4%	99%	91.3%	72%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	92.7%	92.7%	100%	100%	99.8%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC17A8	100%	100%	100%	100%	99.8%	Deafness, autosomal dominant 25, 605583
SLC17A9	99.9%	97.7%	100%	100%	99.5%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC18A2	99.6%	96.7%	100%	100%	99.8%	Parkinsonism-dystonia, infantile, 2, 618049
SLC18A3	100%	100%	100%	100%	98.8%	Myasthenic syndrome, congenital, 21, presynaptic, 617239
SLC19A1	97.7%	97.4%	100%	99.9%	98.9%	Immunodeficiency 114, folate-responsive, 620603;?Megaloblastic anemia, folate-responsive, 601775
SLC19A2	100%	100%	100%	100%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC19A3	99.9%	98.9%	100%	100%	99.7%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC1A1	100%	100%	100%	99.9%	99.4%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232
SLC1A2	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 41, 617105
SLC1A3	97.1%	94.3%	100%	100%	99.6%	Episodic ataxia, type 6, 612656
SLC1A4	100%	100%	100%	99.9%	99.3%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100%	99.5%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 1, 213600
SLC22A12	100%	100%	100%	99.9%	98.7%	Hypouricemia, renal, 220150
SLC22A4	100%	100%	100%	100%	99.5%	{Rheumatoid arthritis, susceptibility to}, 180300
SLC22A5	97.1%	97.1%	100%	100%	99.5%	Carnitine deficiency, systemic primary, 212140
SLC24A1	100%	100%	100%	100%	99.4%	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830
SLC24A4	100%	100%	100%	100%	99.3%	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC24A5	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750
SLC25A1	100%	100%	100%	99.9%	98.8%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A10	99.7%	97.6%	100%	100%	99.2%	?Mitochondrial DNA depletion syndrome 19, 618972
SLC25A11	100%	100%	100%	100%	99.3%	Pheochromocytoma/paraganglioma syndrome 6, 618464
SLC25A12	90.5%	89.5%	100%	100%	99.9%	Developmental and epileptic encephalopathy 39, 612949
SLC25A13	91.7%	91.7%	100%	100%	99.6%	Citrullinemia, type II, neonatal-onset, 605814;Citrullinemia, adult-onset type II, 603471
SLC25A15	100%	100%	100%	100%	99.8%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A19	100%	100%	100%	100%	99.8%	Microcephaly, Amish type, 607196;Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710
SLC25A20	100%	100%	100%	100%	99.4%	Carnitine-acylcarnitine translocase deficiency, 212138
SLC25A21	100%	100%	100%	100%	99.9%	?Mitochondrial DNA depletion syndrome 18, 618811

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC25A22	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99.4%	99.4%	99.7%	99.1%	97.9%	Fontaine progeroid syndrome, 612289
SLC25A26	95.5%	95.5%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 28, 616794
SLC25A3	100%	100%	100%	100%	99.6%	Mitochondrial phosphate carrier deficiency, 610773
SLC25A32	100%	100%	100%	100%	99.7%	?Exercise intolerance, riboflavin-responsive, 616839
SLC25A36	95%	95%	100%	100%	99.4%	Hyperinsulinemic hypoglycemia, familial, 8, 620211
SLC25A38	90.6%	90.6%	100%	100%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC25A4	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283;Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184
SLC25A42	100%	100%	100%	100%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC26A1	100%	100%	100%	100%	99.5%	?Hypersulfaturia, 620372;?Nephrolithiasis, calcium oxalate, 1, 167030
SLC26A2	100%	100%	100%	100%	99.8%	Epiphyseal dysplasia, multiple, 4, 226900;De la Chapelle dysplasia, 256050;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis IB, 600972;Atelosteogenesis, type II, 256050
SLC26A3	100%	100%	100%	100%	99.9%	Diarrhea 1, secretory chloride, congenital, 214700
SLC26A4	100%	100%	100%	100%	99.8%	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791;Pendred syndrome, 274600
SLC26A5	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 61, 613865
SLC26A8	100%	100%	100%	100%	99.5%	Spermatogenic failure 3, 606766
SLC27A3	100%	100%	100%	99.9%	99%	
SLC27A4	100%	100%	100%	100%	99.6%	Ichthyosis prematurity syndrome, 608649
SLC27A5	100%	100%	100%	100%	99.5%	
SLC28A1	100%	100%	100%	100%	99.6%	[Uridine-cytidineuria], 618477

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC29A3	100%	100%	100%	100%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A1	100%	100%	100%	100%	99.3%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC2A10	94.7%	94.7%	100%	100%	99.8%	Arterial tortuosity syndrome, 208050
SLC2A2	100%	100%	100%	100%	99.8%	Fanconi-Bickel syndrome, 227810;{Diabetes mellitus, noninsulin-dependent}, 125853
SLC2A9	100%	100%	100%	100%	99.7%	{Uric acid concentration, serum, QTL 2}, 612076;Hypouricemia, renal, 2, 612076
SLC30A10	100%	100%	100%	100%	99.3%	Hypermanganesemia with dystonia 1, 613280
SLC30A2	99.9%	97.9%	100%	100%	99.6%	Zinc deficiency, transient neonatal, 608118
SLC30A5	100%	100%	100%	100%	99.5%	
SLC30A7	100%	100%	100%	100%	99.9%	Ziegler-Huang syndrome, 620501
SLC30A9	100%	100%	100%	100%	99.9%	Birk-Landau-Perez syndrome, 617595
SLC31A1	100%	100%	100%	99.9%	99.5%	Neurodegeneration and seizures due to copper transport defect, 620306

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC32A1	100%	100%	100%	100%	99.8%	Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC34A1	100%	100%	100%	99.9%	99.4%	?Fanconi renotubular syndrome 2, 613388;Hypercalcemia, infantile, 2, 616963;Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286
SLC34A2	100%	100%	100%	100%	99.5%	Pulmonary alveolar microlithiasis, 265100
SLC34A3	100%	100%	100%	99.8%	98.6%	Hypophosphatemic rickets with hypercalciuria, 241530
SLC35A1	94.9%	94.9%	100%	100%	100%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100%	100%	98.9%	87.9%	68.3%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	91%	87.6%	100%	100%	99.8%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100%	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC35D1	100%	100%	100%	100%	99.8%	Schneckenbecken dysplasia, 269250

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC36A2	100%	100%	100%	100%	99.8%	[Iminoglycinuria], 242600;[Hyperglycinuria], 138500
SLC37A3	100%	100%	100%	100%	99.8%	
SLC37A4	100%	100%	100%	100%	99.8%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC38A3	96.3%	96.3%	100%	100%	99.4%	Developmental and epileptic encephalopathy 102, 619881
SLC38A8	100%	100%	100%	100%	99.5%	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218
SLC39A12	100%	100%	100%	100%	99.8%	
SLC39A13	99.8%	97.5%	100%	100%	99.5%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A14	94%	94%	100%	100%	99.5%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A4	100%	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100
SLC39A5	100%	100%	100%	100%	99.3%	Myopia 24, autosomal dominant, 615946
SLC39A7	100%	100%	100%	100%	99.6%	Agammaglobulinemia 9, autosomal recessive, 619693
SLC39A8	77.5%	77.4%	100%	100%	100%	Congenital disorder of glycosylation, type IIn, 616721
SLC3A1	96.2%	96.2%	100%	100%	99.8%	Cystinuria, 220100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC40A1	100%	100%	100%	100%	99.9%	Hemochromatosis, type 4, 606069
SLC41A1	100%	100%	100%	100%	99.8%	?Nephronophthisis-like nephropathy 2, 619468
SLC44A1	97.5%	97.5%	100%	100%	99.8%	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868
SLC44A4	100%	100%	100%	99.9%	99.2%	?Deafness, autosomal dominant 72, 617606
SLC45A1	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC45A2	100%	100%	100%	100%	99.6%	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240
SLC46A1	100%	100%	99.9%	99.7%	99%	Folate malabsorption, hereditary, 229050
SLC4A1	100%	100%	100%	100%	99.3%	[Blood group, Swann], 601550;[Blood group, Wright], 112050;Distal renal tubular acidosis 1, 179800;[Blood group, Waldner], 112010;Spherocytosis, type 4, 612653;[Blood group, Froese], 601551;Distal renal tubular acidosis 4 with hemolytic anemia, 611590;{Malaria, resistance to}, 611162;Cryohydrocytosis, 185020;Ovalocytosis, SA type, 166900;[Blood group, Diego], 110500

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC4A10	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746
SLC4A11	100%	100%	100%	100%	99.6%	Corneal endothelial dystrophy, autosomal recessive, 217700;Corneal dystrophy, Fuchs endothelial, 4, 613268;Corneal endothelial dystrophy and perceptive deafness, 217400
SLC4A2	100%	100%	100%	100%	99.4%	?Osteopetrosis, autosomal recessive 9, 620366
SLC4A3	100%	100%	100%	100%	99.6%	Short QT syndrome 7, 620231
SLC4A4	95%	95%	100%	100%	99.9%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC4A7	100%	100%	100%	100%	99.8%	
SLC51A	96.2%	96.2%	100%	100%	99.6%	?Cholestasis, progressive familial intrahepatic, 6, 619484
SLC51B	100%	100%	100%	100%	99.4%	?Bile acid malabsorption, primary, 2, 619481
SLC52A1	100%	100%	100%	100%	99.9%	Riboflavin deficiency, 615026
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC5A1	100%	100%	100%	100%	99.4%	Glucose/galactose malabsorption, 606824
SLC5A2	100%	100%	100%	99.9%	99.4%	Renal glucosuria, 233100

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC5A5	100%	100%	100%	100%	99.4%	Thyroid dyshormonogenesis 1, 274400
SLC5A6	100%	100%	100%	100%	99.5%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903
SLC5A7	100%	100%	100%	99.9%	99.4%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC66A1	100%	100%	100%	100%	99.6%	
SLC67A1	87.6%	86.2%	100%	100%	98.9%	Breast cancer, somatic, 114480;Lung cancer, somatic, 211980;Rhabdomyosarcoma, somatic, 268210
SLC6A1	100%	100%	100%	99.9%	99.3%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC6A2	100%	100%	100%	99.9%	99.1%	?Orthostatic intolerance, 604715
SLC6A20	99.9%	98.6%	100%	99.9%	99.5%	
SLC6A3	93.9%	92.4%	100%	100%	99.6%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SLC6A5	100%	100%	100%	100%	99.6%	Hyperekplexia 3, 614618

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLC6A6	100%	100%	100%	100%	99.4%	Hypotaurinemic retinal degeneration and cardiomyopathy, 145350
SLC6A8	100%	99.9%	98.4%	88.1%	71.2%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100%	100%	100%	100%	99.2%	Glycine encephalopathy with normal serum glycine, 617301;{Scoliosis, isolated, susceptibility to, 6}, 621428
SLC7A14	100%	100%	100%	99.9%	99.6%	Retinitis pigmentosa 68, 615725
SLC7A6OS	87%	87%	100%	100%	99.6%	Epilepsy, progressive myoclonic, 12, 619191
SLC7A7	100%	100%	100%	100%	98.8%	Lysinuric protein intolerance, 222700
SLC7A9	100%	100%	100%	100%	99.5%	Cystinuria, 220100
SLC8B1	100%	100%	100%	100%	99.6%	
SLC9A1	100%	100%	100%	99.9%	99.1%	Lichtenstein-Knorr syndrome, 616291
SLC9A3	100%	100%	100%	99.7%	98.5%	Diarrhea 8, secretory sodium, congenital, 616868
SLC9A6	100%	100%	99.1%	90.8%	73.1%	Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairment, 301142;Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	100%	100%	99%	89%	70.1%	Intellectual developmental disorder, X-linked 108, 301024

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SLCO1B1	100%	100%	100%	99.9%	99.6%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO1B3	100%	100%	100%	100%	99.9%	Hyperbilirubinemia, Rotor type, digenic, 237450
SLCO2A1	100%	100%	100%	100%	99.3%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLCO5A1	100%	100%	100%	100%	99.9%	
SLF2	100%	100%	100%	100%	99.8%	Atelis syndrome 1, 620184
SLFN14	100%	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 20, 616913
SLIRP	85.2%	84.3%	100%	100%	99.3%	
SLIT3	100%	100%	100%	100%	99.5%	
SLITRK1	100%	100%	100%	100%	99.8%	Tourette syndrome, 137580;?Trichotillomania, 613229
SLITRK2	100%	100%	99.2%	90.7%	71.1%	Intellectual developmental disorder, X-linked 111, 301107
SLITRK6	100%	100%	100%	100%	99.7%	Deafness and myopia, 221200
SLMAP	100%	100%	100%	100%	99.8%	
SLURP1	100%	100%	100%	100%	99.2%	Meleda disease, 248300
SLX4	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group P, 613951
SMAD1	100%	100%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMAD2	86.7%	86.7%	100%	100%	99.8%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	100%	100%	99.4%	Loeys-Dietz syndrome 3, 613795
SMAD4	95.4%	95.4%	100%	100%	99.8%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD5	100%	100%	100%	100%	99.9%	
SMAD6	100%	100%	100%	99.8%	98.1%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
SMAD9	100%	100%	100%	100%	99.1%	Pulmonary hypertension, primary, 2, 615342
SMARCA1	98%	98%	99.2%	91.5%	73.1%	
SMARCA2	98%	97.9%	100%	100%	99.6%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100%	100%	100%	99.9%	99.3%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCA5	98.4%	98.4%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMARCAD1	100%	100%	100%	100%	99.8%	Basan syndrome, 129200;Huriez syndrome, 181600;Adermatoglyphia, 136000
SMARCAL1	95.6%	95.6%	100%	100%	99.7%	Schimke immunoosseous dysplasia, 242900
SMARCB1	95.2%	95.2%	100%	100%	99.7%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	94.2%	94.2%	100%	99.9%	99.3%	Coffin-Siris syndrome 8, 618362
SMARCD1	100%	100%	100%	100%	99.5%	Coffin-Siris syndrome 11, 618779
SMARCD2	100%	100%	100%	99.9%	99%	Specific granule deficiency 2, 617475
SMARCE1	100%	100%	100%	100%	99.8%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SMC1A	100%	100%	99.1%	88.6%	70.4%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	100%	100%	99.9%	Cornelia de Lange syndrome 3, 610759
SMC5	97.3%	97.3%	100%	100%	99.8%	Atelis syndrome 2, 620185
SMCHD1	100%	100%	100%	100%	99.8%	Facioscapulohumeral muscular dystrophy 2, digenic, 158901;Bosma arhinia microphthalmia syndrome, 603457
SMDT1	100%	100%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMG8	100%	100%	100%	100%	99.7%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100%	100%	100%	100%	99.6%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995
SMN1	93.9%	93.9%	100%	99.5%	98.3%	Spinal muscular atrophy-2, 253550;Spinal muscular atrophy-4, 271150;Spinal muscular atrophy-3, 253400;Spinal muscular atrophy-1, 253300
SMO	100%	100%	100%	100%	99.7%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC1	96%	96%	100%	100%	99.7%	Microphthalmia with limb anomalies, 206920
SMOC2	100%	100%	100%	100%	99.6%	Dentin dysplasia, type IA, 125400
SMPD1	100%	100%	100%	100%	99.6%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SMPD4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMPX	100%	100%	98.9%	89.7%	69.6%	Myopathy, distal, 7, adult-onset, X-linked, 301075;Deafness, X-linked 4, 300066

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMS	94.4%	94.2%	99.1%	91.9%	73.3%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAI2	100%	100%	100%	100%	99.7%	
SNAP25	89.5%	89.5%	100%	100%	99.7%	Developmental and epileptic encephalopathy 117, 616330
SNAP29	100%	100%	100%	99.7%	99.3%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNAPC4	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515
SNAPIN	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with structural brain abnormalities and craniofacial abnormalities, 621393
SNCA	100%	100%	100%	100%	99.9%	Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543
SNCB	70.2%	70.2%	100%	100%	99.3%	Dementia, Lewy body, 127750
SNF8	94.6%	94.6%	100%	100%	99.6%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784
SNIP1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SNORA31						{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 10}, 619396
SNORD118						Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRNP200	100%	100%	100%	100%	99.4%	Retinitis pigmentosa 33, 610359
SNRPB	90.5%	82.4%	100%	100%	99.7%	Cerebrocostomandibular syndrome, 117650
SNRPE	100%	100%	100%	100%	99.9%	Hypotrichosis 11, 615059
SNRPN	100%	100%	100%	100%	99.9%	
SNTA1	92.9%	92.9%	100%	100%	99.3%	Long QT syndrome 12, 612955
SNUPN	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 29, 620793
SNX10	89.3%	89.3%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 8, 615085
SNX14	95%	95%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100%	100%	100%	100%	99.5%	
SOBP	100%	100%	100%	99.8%	97%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671
SOCS1	100%	100%	100%	100%	99.2%	Autoinflammatory syndrome, familial, with or without immunodeficiency, 619375
SOCS4	100%	100%	100%	100%	100%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SOD2	100%	100%	100%	100%	99.8%	{Microvascular complications of diabetes 6}, 612634
SOHLH1	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SON	100%	100%	100%	100%	99.7%	ZTTK syndrome, 617140
SORD	83%	78.5%	98%	95.9%	93.1%	Neuronopathy, distal hereditary motor, autosomal recessive 8, 618912
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	95%	95%	100%	100%	99.5%	Noonan syndrome 9, 616559
SOST	100%	100%	100%	99.9%	98.7%	Sclerosteosis 1, 269500;Craniodiaphyseal dysplasia, autosomal dominant, 122860
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100%	100%	100%	99.7%	96%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SOX17	100%	100%	100%	100%	98.6%	Vesicoureteral reflux 3, 613674;Pulmonary hypertension, primary, 7, 621248
SOX18	100%	100%	100%	99.5%	98%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	100%	100%	100%	99.9%	97.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	100%	100%	96.1%	81.1%	61.5%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX30	100%	100%	100%	100%	99.3%	
SOX4	100%	100%	100%	99.7%	97.5%	Intellectual developmental disorder with speech delay and dysmorphic facies, 618506
SOX5	100%	100%	100%	100%	99.8%	Lamb-Shaffer syndrome, 616803
SOX6	100%	100%	100%	100%	99.6%	Tolchin-Le Caignec syndrome, 618971
SOX7	100%	100%	100%	100%	98.8%	
SOX9	100%	99.5%	100%	99.9%	97.9%	Campomelic dysplasia with autosomal sex reversal, 114290;46XY sex reversal 10, 616425;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290;46XX sex reversal 2, 278850

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SP110	100%	100%	100%	100%	99.8%	{Mycobacterium tuberculosis, susceptibility to}, 607948;Hepatic venoocclusive disease with immunodeficiency, 235550
SP6	100%	100%	100%	99.8%	97.9%	Amelogenesis imperfecta, type IK, 620104
SP7	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XII, 613849
SP9	100%	100%	100%	99.9%	98.4%	
SPACA1	100%	100%	100%	100%	99.9%	?Spermatogenic failure 85, 620490
SPAG1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 28, 615505
SPAG17	100%	100%	100%	100%	99.9%	?Spermatogenic failure 55, 619380
SPAG6	92.8%	92.8%	100%	100%	99.7%	
SPAG9	100%	100%	100%	100%	99.5%	
SPARC	100%	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVII, 616507
SPARCL1	100%	100%	100%	100%	99.9%	
SPART	95.8%	95.8%	100%	100%	99.7%	Troyer syndrome, 275900
SPAST	94.7%	94.6%	100%	100%	99.5%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA16	100%	100%	100%	100%	99.7%	?Spermatogenic failure 6, 102530
SPATA22	100%	100%	100%	100%	99.6%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SPATA7	100%	100%	100%	100%	99.9%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232
SPDYC	100%	100%	100%	99.9%	99.2%	
SPECC1L	94.6%	94.6%	100%	100%	99.7%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPEF2	100%	100%	100%	100%	99.8%	Spermatogenic failure 43, 618751
SPEG	100%	100%	100%	99.9%	98.7%	Centronuclear myopathy 5, 615959
SPEM2	100%	100%	100%	100%	99.5%	
SPEN	100%	100%	100%	100%	99.6%	Radio-Tartaglia syndrome, 619312
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100%	100%	100%	100%	99.7%	Mast syndrome, 248900
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPI1	100%	100%	100%	100%	99.1%	Agammaglobulinemia 10, autosomal dominant, 619707
SPIDR	100%	100%	100%	100%	99.7%	Ovarian dysgenesis 9, 619665
SPIN4	100%	100%	98.3%	87.2%	69.3%	?Lui-Jee-Baron syndrome, 301114

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SPINK1	100%	100%	100%	100%	100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
SPINK2	100%	100%	100%	100%	99.3%	?Spermatogenic failure 29, 618091
SPINK5	100%	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPINT2	100%	100%	100%	100%	99.2%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPNS2	100%	100%	100%	99.8%	99%	?Deafness, autosomal recessive 115, 618457
SPO11	100%	100%	100%	100%	99.8%	
SPOCK1	100%	100%	100%	100%	99.2%	
SPOP	96.4%	96.4%	100%	100%	99.8%	Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829
SPOUT1	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with poor growth, seizures, and brain abnormalities, 621154
SPP2	100%	100%	100%	100%	100%	
SPPL2A	96.2%	96.2%	100%	100%	99.7%	Immunodeficiency 86, mycobacteriosis, 619549
SPR	100%	100%	100%	100%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	97.6%	97.6%	100%	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	100%	100%	99.8%	Noonan syndrome 14, 619745

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SPRTN	100%	100%	100%	100%	99.6%	Ruijs-Aalfs progeroid syndrome, 616200
SPRY4	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SPTA1	100%	100%	100%	100%	99.7%	Spherocytosis, type 3, 270970;Elliptocytosis-2, 130600;Pyropoikilocytosis, 266140
SPTAN1	99.2%	98.8%	100%	100%	99.5%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528
SPTB	100%	100%	100%	100%	99.4%	Anemia, neonatal hemolytic, fatal or near-fatal, 617948;Elliptocytosis-3, 617948;Spherocytosis, type 2, 616649
SPTBN1	100%	100%	100%	100%	99.7%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100%	100%	100%	99.9%	99.3%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	100%	99.9%	98.7%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SPTLC1	84.4%	83.8%	100%	100%	99.7%	Amyotrophic lateral sclerosis 27, juvenile, 620285;Neuropathy, hereditary sensory and autonomic, type IA, 162400
SPTLC2	89%	89%	100%	100%	99.7%	Neuropathy, hereditary sensory and autonomic, type IC, 613640
SPTLC3	91.9%	91.9%	100%	100%	99.9%	
SPTSSA	100%	100%	100%	100%	99.9%	Spastic paraplegia 90A, autosomal dominant, 620416;?Spastic paraplegia 90B, autosomal recessive, 620417
SQOR	100%	100%	100%	100%	99.7%	Sulfide:quinone oxidoreductase deficiency, 619221
SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
SRC	96.5%	96.5%	100%	99.9%	99.3%	?Thrombocytopenia 6, 616937;Colon cancer, advanced, somatic, 114500
SRCAP	100%	100%	100%	100%	99.3%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRD5A2	87.5%	87.5%	100%	100%	99.3%	Pseudovaginal perineoscrotal hypospadias, 264600

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SRD5A3	100%	100%	100%	100%	99.9%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SREBF1	100%	100%	100%	99.7%	98.3%	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016;Mucoepithelial dysplasia, hereditary, 158310
SRF	99.7%	97.4%	100%	99.9%	98.9%	
SRI	100%	100%	100%	100%	99.6%	
SRP19	90.5%	90.5%	100%	100%	99.3%	
SRP54	96.3%	96.3%	100%	100%	99.9%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP68	100%	100%	100%	100%	99.7%	?Neutropenia, severe congenital, 10, autosomal recessive, 620534
SRP72	100%	100%	100%	100%	99.9%	Bone marrow failure syndrome 1, 614675
SRPK3	100%	99.9%	97.7%	87.1%	69.3%	Intellectual developmental disorder, X-linked 114, 301134
SRPRA	100%	100%	100%	100%	99.7%	
SRPX2	100%	100%	99.1%	89.6%	71.9%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM1	100%	100%	100%	100%	99.3%	
SRRM2	100%	100%	100%	99.9%	99.1%	Intellectual developmental disorder, autosomal dominant 72, 620439
SRRM4	100%	100%	100%	100%	99.1%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SRSF1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489
SRY	50%	50%	48.1%	37.3%	22.5%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
SSBP1	100%	100%	100%	100%	100%	Optic atrophy 13 with retinal and foveal abnormalities, 165510
SSPOP						
SSR3	100%	100%	100%	100%	99.4%	
SSR4	100%	100%	98.9%	87.6%	65.5%	Congenital disorder of glycosylation, type Iy, 300934
SSTR5	100%	100%	100%	99.9%	98.6%	
SSX1	100%	100%	99.1%	90.9%	74.2%	Spermatogenic failure, X-linked, 5, 301099;Sarcoma, synovial, somatic, 300813
SSX2	100%	100%	99.7%	93.2%	77.6%	
ST14	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL3	89.5%	88.2%	100%	100%	99.5%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.4%	98.4%	100%	100%	99.6%	Salt and pepper developmental regression syndrome, 609056
STAB2	100%	100%	100%	100%	99.8%	
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

STAG1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	100%	100%	99.3%	91.5%	74%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STAG3	100%	100%	100%	100%	99.3%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261
STAR	100%	100%	100%	100%	99.4%	Lipoid adrenal hyperplasia, 201710
STARD7	91.8%	91.8%	100%	100%	99.1%	Epilepsy, familial adult myoclonic, 2, 607876
STAT1	96%	96%	100%	100%	99.8%	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162;Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892;Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796
STAT2	100%	100%	100%	100%	99.5%	Pseudo-TORCH syndrome 3, 618886;Immunodeficiency 44, 616636
STAT3	100%	100%	100%	100%	99.6%	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

STAT4	93.2%	93.2%	100%	100%	99.7%	Disabling pansclerotic morphea of childhood, 620443;{Systemic lupus erythematosus, susceptibility to, 11}, 612253
STAT5B	100%	100%	100%	100%	99.5%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STAT6	100%	100%	100%	100%	99.4%	Hyper-IgE syndrome 6, autosomal dominant, with recurrent infections, 620532
STEAP3	100%	100%	100%	99.9%	99.3%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
STEEP1	100%	100%	99.1%	88%	67.3%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100%	100%	100%	100%	99.8%	Microcephaly 7, primary, autosomal recessive, 612703
STIM1	100%	99.9%	100%	99.9%	99.7%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STING1	100%	100%	100%	100%	99.4%	STING-associated vasculopathy, infantile-onset, 615934
STK11	99.9%	98.5%	100%	99.9%	98.8%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

STK33	94.3%	94.3%	100%	100%	99.8%	?Spermatogenic failure 93, 620849
STK36	100%	100%	100%	100%	99.7%	?Ciliary dyskinesia, primary, 46, 619436
STK4	100%	100%	100%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STN1	87.1%	87.1%	100%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
STOX1	99.4%	98.4%	94%	90.6%	86.6%	Preeclampsia/eclampsia 4, 609404
STRA6	100%	100%	100%	100%	99.3%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STRA8	100%	100%	100%	100%	99.7%	
STRADA	100%	100%	100%	99.9%	99.4%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STRC	100%	100%	100%	99.9%	99.2%	Deafness, autosomal recessive 16, 603720
STS	97.1%	97.1%	98.9%	90.5%	73.5%	Ichthyosis, X-linked, 308100
STT3A	99.9%	98.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Ix, 615597

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

STUB1	100%	100%	100%	100%	99.4%	Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768
STX11	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 4, 603552
STX16	100%	100%	100%	100%	99.6%	Pseudohypoparathyroidism Ib, 603233
STX1A	100%	98.7%	100%	100%	99.2%	
STX1B	100%	100%	100%	99.8%	98.2%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STX2	100%	100%	100%	100%	99.9%	
STX3	100%	100%	100%	100%	99.7%	Retinal dystrophy and microvillus inclusion disease, 619446;Diarrhea 12, with microvillus atrophy, 619445
STX4	100%	100%	100%	99.9%	99%	?Deafness, autosomal recessive 123, 620745
STX5	100%	100%	100%	100%	99.7%	?Congenital disorder of glycosylation, type IIaa, 620454
STXBP1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 4, 612164
STXBP2	100%	100%	100%	99.9%	99.3%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SUCLG1	100%	100%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUCLG2	100%	100%	100%	100%	99.9%	
SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SUGCT	100%	100%	100%	100%	99.7%	Glutaric aciduria III, 231690
SULF1	100%	100%	100%	100%	99.7%	
SULT2B1	100%	100%	100%	100%	98.7%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200
SUMO1	71%	71%	100%	100%	99.9%	?Orofacial cleft 10, 613705
SUN5	100%	100%	100%	100%	99.2%	Spermatogenic failure 16, 617187
SUOX	100%	100%	100%	100%	99.5%	Sulfite oxidase deficiency, 272300
SUPT16H	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SUPT4H1	100%	100%	100%	100%	99.5%	
SUPT7L	100%	100%	100%	100%	99.9%	Fischer-Zirnsak progeroid syndrome, 621130
SUPV3L1	95.3%	95.3%	100%	100%	99.6%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SURF1	100%	100%	100%	100%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	95.6%	95.6%	100%	100%	99.7%	Imagawa-Matsumoto syndrome, 618786
SV2A	99.8%	98.2%	100%	100%	99.6%	Developmental and epileptic encephalopathy 113, 620772
SVBP	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SVIL	100%	100%	100%	100%	99.4%	Myofibrillar myopathy 10, 619040
SWSAP1	100%	100%	100%	100%	99.1%	
SYCE1	100%	100%	100%	100%	99.3%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2	100%	100%	100%	100%	99.8%	Spermatogenic failure 1, 258150
SYCP2L	100%	100%	100%	100%	99.7%	Premature ovarian failure 24, 620840
SYCP3	100%	100%	100%	99.9%	99.6%	Pregnancy loss, recurrent, 4, 270960;Spermatogenic failure 4, 270960
SYK	100%	100%	100%	100%	99.7%	Immunodeficiency 82 with systemic inflammation, 619381
SYN1	100%	100%	98.4%	86.4%	66.5%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNCRIP	92.9%	92.9%	100%	100%	99.9%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SYNE1	100%	100%	100%	100%	99.8%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743
SYNE2	100%	100%	100%	100%	99.8%	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999
SYNE4	100%	99.7%	100%	100%	99.7%	Deafness, autosomal recessive 76, 615540
SYNGAP1	100%	100%	100%	99.9%	99%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100%	100%	100%	100%	99.8%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYP	100%	100%	98.6%	89.9%	71%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	96%	96%	100%	100%	99.9%	Baker-Gordon syndrome, 618218
SYT14	100%	100%	100%	100%	99.7%	?Spinocerebellar ataxia, autosomal recessive 11, 614229
SYT2	100%	100%	100%	100%	99.4%	Myasthenic syndrome, congenital, 7A, presynaptic, and distal motor neuropathy, autosomal dominant, 616040;Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive, 619461

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SZT2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 18, 615476
TAB2	100%	100%	100%	100%	98.7%	Congenital heart defects, nonsyndromic, 2, 614980
TAB3	100%	100%	99.4%	91.3%	73.6%	
TAC3	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACC3	100%	100%	100%	100%	99.6%	
TACO1	100%	100%	100%	99.8%	99.3%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TACR3	100%	100%	100%	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TACSTD2	100%	100%	100%	99.6%	97.6%	Corneal dystrophy, gelatinous drop-like, 204870
TAF1	98.7%	98.7%	98.9%	89%	70.8%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF13	85.1%	77.2%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1A	91.1%	90.8%	100%	100%	99.9%	
TAF1C	100%	99.2%	100%	100%	99.4%	
TAF2	96.3%	96.3%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 40, 615599

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TAF4	92.3%	84.8%	99.9%	99%	94.1%	Intellectual developmental disorder, autosomal dominant 73, 620450
TAF4B	96.2%	96.2%	100%	100%	99.6%	?Spermatogenic failure 13, 615841
TAF6	100%	100%	100%	100%	99.3%	Alazami-Yuan syndrome, 617126
TAF8	85.3%	85.3%	100%	100%	99.3%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TAFAZZIN	100%	100%	98.6%	89.7%	70.1%	Barth syndrome, 302060
TAL1	100%	100%	100%	100%	98.8%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TAL2	100%	100%	100%	100%	99.8%	Leukemia, T-cell acute lymphocytic, somatic, 613065
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
TAMM41	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 56, 620139
TANC2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TAOK1	100%	100%	100%	100%	99.8%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TAOK2	100%	100%	100%	100%	99.2%	
TAP1	99.7%	97.9%	100%	100%	99.7%	MHC class I deficiency 1, 604571
TAP2	97.9%	97.9%	100%	99.9%	99%	MHC class I deficiency 2, 620813
TAPBP	88.8%	88.8%	100%	99.8%	98%	?MHC class I deficiency 3, 620814
TAPT1	100%	100%	100%	100%	99.7%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TARDBP	100%	100%	100%	100%	99.7%	Frontotemporal lobar degeneration, TARDBP-related, 612069;Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TARS1	100%	100%	100%	100%	99.9%	Trichothiodystrophy 7, nonphotosensitive, 618546
TARS2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 21, 615918
TASP1	93.2%	93.2%	100%	100%	99.8%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	100%	100%	99.8%	Tyrosinemia, type II, 276600
TAX1BP3	100%	100%	100%	100%	99.2%	
TBC1D20	92.2%	92.2%	100%	100%	99.7%	Warburg micro syndrome 4, 615663
TBC1D23	96.3%	96.3%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 11, 617695

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TBC1D24	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBC1D2B	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D32	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome IX, 258865;Alsahan-Harris syndrome, 621307;Retinitis pigmentosa 100, 621280
TBC1D7	88.9%	88.9%	100%	100%	99.4%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBC1D8	100%	100%	100%	99.9%	99.3%	
TBC1D8B	100%	100%	99.3%	92.3%	75.5%	Nephrotic syndrome, type 20, 301028
TBCB	77.5%	77.5%	100%	100%	99.4%	Neurodevelopmental disorder with behavioral abnormalities and childhood onset spastic paraplegia, 621382
TBCD	91.2%	90.7%	100%	99.9%	99.2%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TBCE	96.7%	96.6%	100%	100%	99.6%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100%	100%	100%	100%	99.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBK1	100%	100%	100%	100%	99.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900;Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439;Autoinflammation with arthritis and vasculitis, 620880
TBL1X	100%	100%	98.6%	88.7%	70.4%	Hypothyroidism, congenital, nongoitrous, 8, 301033
TBL1XR1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342
TBL1Y	50%	50%	48.1%	37.9%	20.5%	?Deafness, Y-linked 2, 400047
TBP	100%	100%	100%	100%	99.3%	Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600
TBPL2	100%	100%	100%	100%	99.9%	
TBR1	100%	100%	100%	99.9%	98.9%	Intellectual developmental disorder with autism and speech delay, 606053

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TBX1	98.6%	95.7%	100%	99.4%	96.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX15	100%	100%	100%	100%	99.8%	Cousin syndrome, 260660
TBX18	100%	100%	100%	100%	99.2%	Congenital anomalies of kidney and urinary tract 2, 143400
TBX19	100%	100%	100%	100%	99.6%	Adrenocorticotropic hormone deficiency, 201400
TBX2	99.5%	97.7%	100%	100%	99.1%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX20	100%	100%	100%	99.9%	99.4%	Atrial septal defect 4, 611363
TBX21	100%	100%	100%	99.9%	98.7%	Asthma and nasal polyps, 208550;?Immunodeficiency 88, 619630;{Asthma, aspirin-induced, susceptibility to}, 208550
TBX22	100%	100%	99%	91.2%	72.3%	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905
TBX3	100%	100%	100%	100%	99.2%	Ulnar-mammary syndrome, 181450
TBX4	100%	100%	100%	100%	99%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TBX5	100%	100%	100%	100%	99.4%	Holt-Oram syndrome, 142900
TBX6	100%	99.5%	100%	100%	99.3%	Spondylocostal dysostosis 5, 122600
TBXA2R	100%	99.6%	100%	100%	99.5%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	100%	100%	100%	100%	99.8%	Ghosal hematodiaphyseal syndrome, 231095
TBXT	100%	100%	100%	100%	99.6%	Sacral agenesis with vertebral anomalies, 615709;{Neural tube defects, susceptibility to}, 182940
TCAP	100%	100%	100%	100%	99.7%	Cardiomyopathy, hypertrophic, 25, 607487;Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954
TCEAL1	100%	100%	99.7%	86.3%	68.4%	Hijazi-Reis syndrome, 301094
TCF12	96.1%	96.1%	100%	100%	99.7%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCF20	100%	100%	100%	99.9%	99.1%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF3	100%	100%	100%	100%	99.3%	Agammaglobulinemia 8B, autosomal recessive, 619824;Agammaglobulinemia 8A, autosomal dominant, 616941
TCF4	97.1%	97.1%	100%	100%	99.6%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TCF7L2	100%	100%	100%	99.9%	98.6%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCHH	100%	100%	100%	99.7%	97.5%	?Uncombable hair syndrome 3, 617252
TCIRG1	100%	100%	100%	100%	99.7%	Osteopetrosis, autosomal recessive 1, 259700
TCN2	91.8%	87.7%	100%	100%	99.9%	Transcobalamin II deficiency, 275350
TCOF1	97.9%	97.9%	100%	100%	99.6%	Treacher Collins syndrome 1, 154500
TCP1	100%	100%	100%	99.9%	99.7%	Intellectual developmental disorder with polymicrogyria and seizures, 621021
TCTN1	92.6%	90.8%	100%	100%	99.5%	Joubert syndrome 13, 614173
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDP1	87.9%	87.5%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TDRD12	100%	100%	100%	100%	99.7%	
TDRD7	100%	100%	100%	100%	99.8%	Cataract 36, 613887
TDRD9	100%	100%	100%	100%	99.5%	?Spermatogenic failure 30, 618110
TDRKH	100%	100%	100%	99.6%	98.6%	
TEAD1	100%	100%	100%	99.9%	99.5%	Sveinsson chorioretinal atrophy, 108985

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TECPR2	100%	100%	100%	99.9%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	87.3%	87.3%	100%	100%	98.9%	Intellectual developmental disorder, autosomal recessive 14, 614020
TECRL	78%	78%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021
TECTA	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 8/12, 601543;Deafness, autosomal recessive 21, 603629
TEFM	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 58, 620451
TEK	100%	100%	100%	100%	99.9%	Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272
TEKT3	100%	100%	100%	100%	99.7%	Spermatogenic failure 81, 620277
TELO2	94.4%	94.3%	100%	99.9%	99.4%	You-Hoover-Fong syndrome, 616954
TENM1	100%	100%	99.2%	91%	72.1%	
TENM3	99.6%	99.6%	100%	100%	99.6%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145
TENM4	100%	100%	100%	100%	99.6%	Essential tremor, hereditary, 5, 616736
TENT5A	100%	100%	100%	100%	99.4%	Osteogenesis imperfecta, type XVIII, 617952
TENT5D	100%	100%	99.7%	93.2%	75.2%	
TERB1	100%	100%	100%	100%	99.8%	Spermatogenic failure 60, 619646

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TERB2	100%	100%	100%	100%	99.4%	?Spermatogenic failure 59, 619645
TERC						Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	97%	95.4%	100%	99.9%	99.3%	
TERT	99.6%	97.7%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TES	100%	100%	100%	100%	99.9%	
TET2	100%	100%	100%	100%	99.9%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
TET3	100%	100%	100%	100%	98.9%	Beck-Fahrner syndrome, 618798
TEX11	97.1%	97.1%	99.5%	91.9%	73.8%	Spermatogenic failure, X-linked 2, 309120
TEX14	100%	100%	100%	100%	99.7%	Spermatogenic failure 23, 617707
TEX15	100%	100%	100%	100%	99.7%	Spermatogenic failure 25, 617960
TF	100%	100%	100%	100%	99.5%	Atransferrinemia, 209300
TFAM	100%	100%	100%	99.9%	99.4%	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
TFAP2B	100%	100%	100%	100%	99.4%	Patent ductus arteriosus 2, 617035;Char syndrome, 169100
TFB2M	100%	100%	100%	100%	99.8%	
TFDP3	100%	100%	98.6%	86.1%	66%	
TFE3	100%	99.9%	98.4%	87.4%	67.8%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854
TFG	100%	100%	100%	100%	99.4%	?Spastic paraplegia 57, autosomal recessive, 615658;Hereditary motor and sensory neuropathy, Okinawa type, 604484
TFPT	100%	100%	100%	100%	99%	
TFR2	95.8%	94.2%	100%	100%	99%	Hemochromatosis, type 3, 604250
TFRC	94.7%	94.7%	100%	99.9%	99.4%	Immunodeficiency 46, 616740
TG	100%	100%	100%	99.9%	99.2%	{Autoimmune thyroid disease, susceptibility to, 3}, 608175;Thyroid dyshormonogenesis 3, 274700
TGDS	100%	100%	100%	100%	99.9%	Catel-Manzke syndrome, 616145

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TGFB1	100%	100%	100%	99.9%	98.5%	Inflammatory bowel disease, immunodeficiency, and encephalopathy, 618213;{Cystic fibrosis lung disease, modifier of}, 219700;Camurati-Engelmann disease 1, 131300
TGFB2	100%	100%	100%	99.8%	99.4%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	88.3%	88.3%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBI	100%	100%	100%	100%	99.7%	Corneal dystrophy, Avellino type, 607541;Corneal dystrophy, Reis-Bucklers type, 608470;Corneal dystrophy, Thiel-Behnke type, 602082;Corneal dystrophy, Groenouw type I, 121900;Corneal dystrophy, epithelial basement membrane, 121820;Corneal dystrophy, lattice type I, 122200;Corneal dystrophy, lattice type IIIA, 608471
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	99.6%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TGIF1	100%	100%	100%	100%	99.9%	Holoprosencephaly 4, 142946
TGM1	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100%	100%	100%	100%	99.7%	?Uncombable hair syndrome 2, 617251
TGM5	100%	100%	100%	100%	99.3%	Peeling skin syndrome 2, 609796
TGM6	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 35, 613908
TH	100%	99.8%	100%	100%	99.2%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	100%	100%	99.8%	Dystonia 6, torsion, 602629
THAP11	100%	100%	100%	100%	98.6%	?Methylmalonic aciduria and homocystinuria, cblL type, 620940;Spinocerebellar ataxia 51, 620947
THBD	100%	100%	100%	99.8%	98.7%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THBS1	100%	100%	100%	100%	99.6%	
THBS2	100%	100%	100%	100%	99.1%	?Ehlers-Danlos syndrome, classic-like, 3, 620865;{Lumbar disc herniation, susceptibility to}, 603932
THBS4	100%	100%	100%	100%	99.7%	
THG1L	100%	100%	100%	99.7%	99.3%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC1	100%	100%	100%	100%	99.8%	?Deafness, autosomal dominant 86, 620280

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

THOC2	100%	100%	99.2%	91.7%	73.7%	Arthrogryposis multiplex congenita 7, X-linked, 301127;Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957
THOC6	100%	100%	100%	100%	99.5%	Beaulieu-Boycott-Innes syndrome, 613680
THPO	100%	100%	100%	100%	99.3%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
THRA	100%	100%	100%	100%	99.4%	Hypothyroidism, congenital, nongoitrous, 6, 614450
THRB	100%	100%	100%	100%	99.9%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
THSD1	100%	100%	100%	100%	99.6%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
THSD4	100%	100%	100%	99.9%	99.5%	Aortic aneurysm, familial thoracic 12, 619825
THUMPD1	100%	100%	100%	99.8%	99.4%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TIA1	92.7%	92.7%	100%	100%	99.8%	Welander distal myopathy, 604454;Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133
TIAM1	98.3%	98.3%	100%	100%	99.5%	Neurodevelopmental disorder with language delay and seizures, 619908
TICAM1	100%	100%	100%	100%	99.6%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 6}, 614850
TIE1	100%	100%	100%	100%	99.7%	Lymphatic malformation 11, 619401
TIMELESS	100%	100%	100%	100%	99.6%	?Advance sleep phase syndrome, familial, 4, 620015
TIMM22	100%	100%	100%	99.8%	98.3%	?Combined oxidative phosphorylation deficiency 43, 618851
TIMM44	100%	100%	100%	99.8%	98.7%	
TIMM50	100%	100%	100%	100%	99.6%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100%	100%	98.2%	87.9%	71.6%	Mohr-Tranebjaerg syndrome, 304700
TIMMDC1	100%	100%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 31, 618251
TIMP3	84%	84%	100%	100%	99.4%	Sorsby fundus dystrophy, 136900
TINF2	100%	100%	100%	100%	99.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TIRAP	100%	100%	100%	100%	99.9%	{Malaria, protection against}, 611162;{Tuberculosis, protection against}, 607948;{Bacteremia, protection against}, 614382
TJP1	100%	100%	100%	100%	99.7%	
TJP2	97.1%	95.8%	100%	100%	99.8%	Hypercholanemia, familial 1, 607748;Cholestasis, progressive familial intrahepatic 4, 615878
TK2	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560;?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069
TKFC	100%	100%	100%	99.9%	99.4%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	89%	89%	100%	100%	99.5%	Short stature, developmental delay, and congenital heart defects, 617044
TKTL1	100%	100%	99.1%	90.8%	70.6%	
TLCD3B	100%	100%	100%	99.8%	99%	Cone-rod dystrophy 22, 619531
TLE6	100%	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 15, 616814
TLK2	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal dominant 57, 618050
TLL1	100%	100%	100%	100%	99.8%	Atrial septal defect 6, 613087

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TLR1	100%	100%	100%	100%	100%	{Leprosy, susceptibility to, 5}, 613223;{Leprosy, protection against}, 613223
TLR3	100%	100%	100%	100%	99.7%	{HIV1 infection, resistance to}, 609423;{Immunodeficiency 83, susceptibility to viral infections}, 613002
TLR4	100%	100%	100%	100%	99.8%	
TLR5	100%	100%	100%	100%	99.5%	{Melioidosis, susceptibility to}, 615557;{Systemic lupus erythematosus, susceptibility to, 1}, 601744;{Systemic lupus erythematosus, resistance to}, 601744;{Legionnaire disease, susceptibility to}, 608556
TLR7	100%	100%	99.3%	92.7%	74.5%	Immunodeficiency 74, COVID19-related, X-linked, 301051;Systemic lupus erythematosus 17, 301080
TLR8	100%	100%	99.2%	91.7%	74.5%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TM2D3	100%	100%	100%	100%	99.6%	Neurocardiorenal malformation syndrome, 621379
TMC1	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 36, 606705;Deafness, autosomal recessive 7, 600974
TMC6	94.9%	94.4%	100%	99.9%	99.1%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TMC8	100%	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TMCO1	72.3%	72.3%	100%	100%	99.6%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMCO3	94.6%	94.6%	100%	99.9%	99.1%	
TMEM106B	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100%	100%	100%	100%	99.2%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM126A	100%	100%	100%	100%	100%	Optic atrophy 7, 612989
TMEM126B	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 29, 618250
TMEM127	100%	100%	100%	100%	99.5%	{Pheochromocytoma, susceptibility to}, 171300
TMEM132E	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 99, 618481
TMEM138	100%	98.8%	100%	99.9%	99.7%	Joubert syndrome 16, 614465
TMEM147	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075
TMEM14C	100%	100%	100%	100%	99.8%	
TMEM151A	100%	100%	100%	100%	98.3%	Episodic kinesigenic dyskinesia 3, 620245
TMEM163	93.9%	93.9%	100%	99.9%	99.1%	Leukodystrophy, hypomyelinating, 25, 620243

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TMEM165	88.9%	88.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM167A	100%	100%	100%	100%	99.7%	
TMEM184B	100%	100%	100%	100%	99.2%	
TMEM186	100%	100%	100%	100%	99.8%	
TMEM216	100%	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM218	100%	100%	100%	100%	99.6%	Joubert syndrome 39, 619562
TMEM222	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	93.5%	93.5%	100%	100%	99.5%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	100%	99.7%	Joubert syndrome 14, 614424
TMEM240	100%	100%	100%	99.7%	97.6%	Spinocerebellar ataxia 21, 607454
TMEM260	94.8%	94.8%	100%	100%	99.7%	Structural heart defects and renal anomalies syndrome, 617478
TMEM38B	89.5%	89.5%	100%	100%	99.9%	Osteogenesis imperfecta, type XIV, 615066
TMEM43	100%	100%	100%	100%	99.5%	Arrhythmogenic right ventricular dysplasia 5, 604400;Auditory neuropathy, autosomal dominant 3, 619832;Emery-Dreifuss muscular dystrophy 7, AD, 614302
TMEM53	100%	100%	100%	100%	99.3%	Craniotubular dysplasia, Ikegawa type, 619727

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TMEM63A	99.7%	97.5%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM63B	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 118, 621250
TMEM63C	100%	100%	100%	100%	99.1%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM65	100%	99.5%	100%	99.7%	98.5%	
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TMEM70	100%	100%	100%	100%	99.3%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	100%	100%	99.6%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMEM98	100%	100%	100%	100%	99.9%	Nanophthalmos 4, 615972
TMIE	100%	100%	100%	99.9%	99.4%	Deafness, autosomal recessive 6, 600971
TMLHE	94.8%	94.8%	99.6%	94.8%	79.1%	{Autism, susceptibility to, X-linked 6}, 300872
TMPO	100%	100%	100%	100%	99.8%	
TMPRSS15	100%	100%	100%	100%	99.7%	Enterokinase deficiency, 226200
TMPRSS3	100%	100%	100%	100%	99.5%	Deafness, autosomal recessive 8/10, 601072

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TMPRSS6	100%	99.9%	100%	100%	99.5%	Iron-refractory iron deficiency anemia, 206200
TMTC2	97.3%	97.1%	100%	100%	99.7%	
TMTC3	100%	100%	100%	100%	99.7%	Lissencephaly 8, 617255
TMTC4	100%	100%	100%	100%	99.9%	?Deafness, autosomal recessive 122, 620714
TMX2	99.8%	97%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNC	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 56, 615629
TNF	100%	100%	100%	100%	99.5%	{Migraine without aura, susceptibility to}, 157300;{Dementia, vascular, susceptibility to};?Immunodeficiency 127, 620977;{Asthma, susceptibility to}, 600807;{Septic shock, susceptibility to};{Malaria, cerebral, susceptibility to}, 611162
TNFAIP3	100%	100%	100%	100%	99.8%	Autoinflammatory syndrome, familial, Behcet-like 1, 616744
TNFRSF10B	100%	100%	100%	100%	99.4%	Squamous cell carcinoma, head and neck, 275355
TNFRSF11A	100%	100%	100%	100%	99.5%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFRSF11B	89.8%	89.8%	100%	100%	99.9%	Paget disease of bone 5, juvenile-onset, 239000

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TNFRSF13B	100%	100%	100%	100%	99%	Immunodeficiency, common variable, 2, 240500;Immunoglobulin A deficiency 2, 609529
TNFRSF13C	100%	100%	100%	99.9%	98.5%	Immunodeficiency, common variable, 4, 613494
TNFRSF1A	100%	100%	100%	100%	99.6%	{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680
TNFRSF4	100%	99.8%	100%	99.7%	98.9%	?Immunodeficiency 16, 615593
TNFRSF9	100%	100%	100%	100%	99.9%	Immunodeficiency 109 with lymphoproliferation, 620282
TNFSF11	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 2, 259710
TNFSF12	100%	100%	100%	99.9%	99.3%	
TNFSF13	100%	100%	100%	100%	99.6%	
TNIK	98.6%	98.6%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNNC1	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNC2	100%	100%	100%	99.9%	99.1%	Congenital myopathy 15, 620161
TNNI2	100%	100%	100%	100%	99.1%	Arthrogryposis, distal, type 2B1, 601680

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TNNI3	100%	100%	100%	100%	98.2%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	100%	100%	99.8%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT1	99.9%	97.6%	100%	99.9%	98.5%	Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386
TNNT2	100%	100%	100%	100%	99.7%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494
TNNT3	100%	100%	100%	100%	99.5%	Arthrogryposis, distal, type 2B2, 618435
TNPO2	100%	100%	100%	99.9%	99.2%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNPO3	97.1%	97.1%	100%	100%	99.9%	Muscular dystrophy, limb-girdle, autosomal dominant 2, 608423

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TNR	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6A	100%	100%	100%	100%	99.7%	?Epilepsy, familial adult myoclonic, 6, 618074
TNRC6B	100%	100%	100%	100%	99.6%	Global developmental delay with speech and behavioral abnormalities, 619243
TNS1	100%	100%	100%	100%	99.4%	
TNS2	100%	100%	100%	99.9%	98.5%	
TNXB	100%	100%	100%	99.9%	98.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TOE1	100%	100%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOM1	65.9%	64.9%	100%	100%	99.6%	?Immunodeficiency 85 and autoimmunity, 619510
TOMM40L	100%	100%	100%	100%	99.3%	
TOMM7	100%	100%	100%	100%	99.6%	Garg-Mishra progeroid syndrome, 620601
TOMM70	100%	100%	100%	100%	99.7%	
TONSL	96.1%	96.1%	100%	99.9%	99.3%	Spondyloepimetaphyseal dysplasia, sponastrime type, 271510
TOP1	100%	100%	100%	100%	99.9%	DNA topoisomerase I, camptothecin-resistant
TOP2A	100%	100%	100%	100%	99.8%	DNA topoisomerase II, resistance to inhibition of, by amsacrine

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TOP2B	100%	100%	100%	100%	99.7%	B-cell immunodeficiency, distal limb anomalies, and urogenital malformations, 609296
TOP3A	95.7%	95%	100%	100%	99.5%	Microcephaly, growth restriction, and increased sister chromatid exchange 2, 618097;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5, 618098
TOP6BL	89.2%	89.1%	100%	100%	99.7%	Hydatidiform mole, recurrent, 4, 618432
TOPORS	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 31, 609923
TOR1A	93.1%	90.7%	100%	99.9%	99.4%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TOR1AIP1	100%	100%	100%	100%	99.8%	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures, 617072

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TP53	95.1%	95.1%	100%	99.9%	99.4%	{Basal cell carcinoma 7}, 614740;{Adrenocortical carcinoma, pediatric}, 202300;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome, 151623;Pancreatic cancer, somatic, 260350;Nasopharyngeal carcinoma, somatic, 607107;{Osteosarcoma}, 259500;{Choroid plexus papilloma}, 260500;{Colorectal cancer}, 114500;{Glioma susceptibility 1}, 137800;Bone marrow failure syndrome 5, 618165
TP53RK	100%	100%	100%	100%	99.8%	Galloway-Mowat syndrome 4, 617730
TP63	100%	100%	100%	100%	99.6%	Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543
TP73	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPCN2	100%	100%	100%	100%	99.5%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TPI1	100%	100%	100%	100%	99.6%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPK1	93.3%	93.3%	100%	100%	99.9%	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458
TPM1	91.3%	91.3%	100%	100%	99.5%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TPM2	100%	100%	100%	100%	99.5%	Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285
TPM3	100%	100%	100%	99.9%	99.8%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284
TPM4	89.4%	88.4%	100%	100%	99%	Bleeding disorder, platelet-type, 25, 620486
TPMT	100%	100%	100%	100%	99.7%	{Thiopurines, poor metabolism of, 1}, 610460
TPO	100%	100%	100%	100%	99.5%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100%	100%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	100%	100%	99.9%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TPR	100%	100%	100%	100%	99.6%	?Intellectual developmental disorder, autosomal recessive 79, 620393
TPRKB	83.2%	81.2%	100%	100%	99.8%	Galloway-Mowat syndrome 5, 617731
TPRN	97.4%	95.3%	100%	99.7%	96.6%	Deafness, autosomal recessive 79, 613307
TRA2B	100%	100%	100%	100%	99.7%	Ramond-Elliott neurodevelopmental syndrome, 621421
TRAC	100%	100%	100%	100%	99.7%	
TRAF3	100%	100%	100%	99.8%	98.9%	Immunodeficiency 132A, 614849;Immunodeficiency 132B, 621096
TRAF3IP1	100%	100%	100%	100%	99.6%	
TRAF3IP2	100%	99.2%	100%	100%	99.7%	?Candidiasis, familial, 8, 615527;{Psoriasis susceptibility 13}, 614070
TRAF6	100%	100%	100%	100%	99.8%	
TRAF7	100%	100%	100%	99.9%	99.2%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100%	100%	100%	100%	99.8%	Seckel syndrome 9, 616777
TRAK1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC10	97.6%	97.6%	100%	99.9%	99.4%	Neurodevelopmental disorder with microcephaly, short stature, and speech delay, 620027
TRAPPC11	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TRAPPC12	86.3%	86.3%	100%	99.9%	99%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC14	100%	100%	100%	100%	99.5%	?Microcephaly 25, primary, autosomal recessive, 618351
TRAPPC2	100%	100%	99.8%	94.1%	77.5%	Spondyloepiphyseal dysplasia tarda, 313400
TRAPPC2L	100%	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100%	100%	100%	99.9%	99.3%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 13, 613192
TRDN	92.7%	92.7%	100%	100%	99.8%	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441
TREH	100%	100%	100%	100%	99.7%	Trehalase deficiency, 612119
TREM2	100%	100%	100%	100%	99.9%	{Alzheimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRH	100%	100%	100%	100%	99.5%	Thyrotropin-releasing hormone deficiency, 275120
TRHR	100%	100%	100%	100%	99.9%	Hypothyroidism, congenital, nongoitrous, 7, 618573
TRIM2	96.1%	96.1%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 2R, 615490
TRIM22	100%	100%	100%	100%	99.7%	
TRIM28	100%	100%	100%	100%	99.4%	Wilms tumor 7, 621332
TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM33	94.6%	94.6%	100%	100%	99.5%	?Developmental dysplasia of the hip 4, 621311
TRIM36	100%	100%	100%	100%	99.8%	?Anencephaly 1, 206500
TRIM37	98.3%	97.6%	100%	100%	99.7%	Mulibrey nanism, 253250
TRIM44	80.6%	80.6%	100%	100%	99.6%	?Aniridia 3, 617142
TRIM63	100%	100%	100%	100%	99.7%	Cardiomyopathy, familial hypertrophic, 31, 621270

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TRIM71	100%	100%	100%	100%	98.9%	Hydrocephalus, congenital, 4, 618667
TRIM8	100%	100%	100%	99.9%	99.2%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99.3%	99.1%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIOBP	100%	100%	100%	99.9%	99.1%	Deafness, autosomal recessive 28, 609823
TRIP11	100%	100%	100%	100%	99.8%	Odontochondrodysplasia 1, 184260;Achondrogenesis, type IA, 200600
TRIP12	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIP13	100%	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 9, 619011;Mosaic variegated aneuploidy syndrome 3, 617598
TRIP4	95.9%	94.9%	100%	100%	99.7%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866
TRIT1	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	100%	100%	99%	Intellectual developmental disorder, autosomal recessive 68, 618302

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TRMT10A	100%	100%	100%	100%	99.7%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT10C	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 30, 616974
TRMT5	100%	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539
TRMU	88%	88%	100%	100%	99.4%	{Deafness, mitochondrial, modifier of}, 580000;Liver failure, transient infantile, 613070
TRNT1	91.9%	91.9%	100%	100%	99.9%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPA1	100%	100%	100%	100%	99.8%	?Episodic pain syndrome, familial, 1, 615040
TRPC3	100%	100%	100%	99.9%	99.5%	?Spinocerebellar ataxia 41, 616410
TRPC6	100%	100%	100%	100%	99.7%	Glomerulosclerosis, focal segmental, 2, 603965
TRPM1	100%	100%	100%	100%	99.8%	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TRPM3	97.8%	97.8%	100%	100%	99.6%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TRPM4	100%	100%	100%	100%	99.4%	Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531
TRPM6	100%	100%	100%	100%	99.8%	Hypomagnesemia 1, intestinal, 602014
TRPM7	100%	100%	100%	100%	99.8%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500
TRPM8	100%	100%	100%	100%	99.7%	
TRPS1	100%	100%	100%	100%	99.8%	Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350
TRPV1	100%	100%	100%	100%	99.3%	
TRPV3	96.6%	96.6%	100%	100%	99.3%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TRPV4	100%	100%	100%	99.9%	99.1%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500
TRPV6	100%	100%	100%	100%	99.2%	Hyperparathyroidism, transient neonatal, 618188
TRRAP	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	100%	100%	99.3%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TSEN15	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100%	100%	99.6%	Pontocerebellar hypoplasia type 2B, 612389
TSEN34	100%	100%	100%	100%	98.8%	?Pontocerebellar hypoplasia type 2C, 612390
TSEN54	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TSFM	91.5%	91.1%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 3, 610505
TSGA10	96.5%	96.5%	100%	100%	99.8%	?Spermatogenic failure 26, 617961
TSHB	100%	100%	100%	100%	99.7%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSHR	100%	100%	100%	100%	99.7%	Hyperthyroidism, familial gestational, 603373;Hyperthyroidism, nonautoimmune, 609152;Thyroid adenoma, hyperfunctioning, somatic, 609152;Hypothyroidism, congenital, nongoitrous, 1, 275200;Thyroid carcinoma with thyrotoxicosis, somatic, 609152
TSHZ1	100%	100%	100%	99.9%	99.4%	Aural atresia, congenital, 607842
TSPAN12	100%	100%	100%	100%	99.7%	Exudative vitreoretinopathy 5, 613310

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TSPAN7	100%	100%	99.2%	90.3%	72.7%	Intellectual developmental disorder, X-linked 58, 300210
TSPEAR	93.9%	93.9%	100%	100%	99.1%	Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hypohidrotic/hair/tooth/ nail type, 618180
TSPOAP1	100%	100%	100%	100%	99.5%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456
TSPYL1	100%	100%	100%	100%	99.1%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TSR2	100%	100%	98.4%	88%	67.7%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TTBK2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 11, 604432
TTC12	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 45, 618801
TTC19	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC21A	100%	100%	100%	100%	99.8%	Spermatogenic failure 37, 618429
TTC21B	98.4%	98.1%	100%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthi sis 12, 613820
TTC29	100%	100%	100%	100%	99.9%	Spermatogenic failure 42, 618745

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TTC5	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC7A	100%	99.8%	100%	100%	99.6%	Gastrointestinal defects and immunodeficiency syndrome, 243150
TTC8	91.4%	91.4%	100%	100%	99.9%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTI1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445
TTI2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 39, 615541
TTLL5	100%	100%	100%	100%	99.7%	Cone-rod dystrophy 19, 615860
TTN	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;?Cardiomyopathy, familial hypertrophic, 9, 613765;Myopathy myofibrillar, 9, with early respiratory failure, 603689
TTPA	87.1%	87.1%	100%	100%	99.7%	Ataxia with isolated vitamin E deficiency, 277460

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TTR	100%	100%	100%	100%	99.5%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680
TTTY5						
TUB	100%	100%	100%	100%	99.5%	?Retinal dystrophy and obesity, 616188
TUBA1A	100%	100%	100%	100%	99.6%	Lissencephaly 3, 611603
TUBA1C	100%	100%	100%	99.9%	99.3%	Oocyte/zygote/embryo maturation arrest 24, 621232
TUBA3D	100%	99.9%	100%	100%	99.6%	Keratoconus 9, 617928
TUBA4A	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 23, 621231;Spastic ataxia 11, autosomal dominant, 621226;Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208;Congenital myopathy 26, 621225
TUBA8	95.3%	95.1%	100%	99.8%	99.2%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840
TUBB	99.8%	98.9%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB1	100%	100%	100%	100%	99.8%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TUBB2A	100%	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	100%	100%	100%	99.4%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100%	100%	100%	100%	99.5%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99.2%	96.6%	100%	100%	99%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBB4B	100%	100%	100%	100%	99.3%	Leber congenital amaurosis with early-onset deafness, 617879
TUBB6	100%	100%	100%	100%	99.5%	?Facial palsy, congenital, with ptosis and velopharyngeal dysfunction, 617732
TUBB8	100%	100%	100%	100%	99.5%	Oocyte/zygote/embryo maturation arrest 2, 616780
TUBG1	100%	100%	100%	100%	99.5%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97%	97%	100%	100%	99.6%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737
TUBGCP4	100%	100%	100%	99.8%	98.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TUBGCP6	100%	100%	100%	99.9%	99.2%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUFM	99.7%	96.7%	100%	99.9%	99.4%	Combined oxidative phosphorylation deficiency 4, 610678
TUFT1	100%	100%	100%	100%	99.4%	Woolly hair-skin fragility syndrome, 620415
TULP1	100%	100%	100%	100%	99.5%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132
TULP3	93.4%	93.4%	100%	100%	99.5%	Hepatorenocardiac degenerative fibrosis, 619902
TUSC3	93.3%	93.3%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	100%	100%	100%	100%	98.5%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWIST2	100%	100%	100%	99.9%	96.6%	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260
TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TXN2	80.8%	80.6%	100%	100%	99.9%	?Combined oxidative phosphorylation deficiency 29, 616811
TXNDC15	100%	98.5%	100%	99.9%	99.5%	Meckel syndrome 14, 619879
TXNIP	100%	100%	100%	100%	99.7%	
TXNL4A	100%	100%	100%	99.7%	99.5%	Burn-McKeown syndrome, 608572
TXNRD2	100%	100%	100%	100%	99.4%	?Glucocorticoid deficiency 5, 617825
TYK2	100%	99.8%	100%	99.9%	98.7%	Immunodeficiency 35, 611521
TYMP	100%	100%	100%	100%	99.4%	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041
TYMS	89.8%	85.5%	100%	100%	99.7%	Dyskeratosis congenita, digenic, 620040
TYR	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100
TYROBP	91.4%	91.2%	100%	99.8%	99.2%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

TYRP1	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
U2AF2	100%	100%	100%	99.9%	99.2%	Developmental delay, dysmorphic facies, and brain anomalies, 620535
UBA1	96.3%	95.6%	99%	90%	71.9%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UBA2	100%	100%	100%	100%	99.6%	ACCES syndrome, 619959
UBA5	94.1%	92.2%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBAP1	96.8%	96.8%	100%	100%	99.5%	Spastic paraplegia 80, autosomal dominant, 618418
UBAP1L	100%	100%	100%	100%	99.5%	Retinal dystrophy, Zeitz-Han type, 621558
UBAP2L	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494
UBB	100%	100%	100%	99.9%	97.7%	
UBE2A	95.6%	91.8%	99%	90.8%	72.2%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

UBE2T	92.6%	92.6%	100%	100%	99.7%	Fanconi anemia, complementation group T, 616435
UBE3A	100%	100%	100%	100%	99.7%	Angelman syndrome, 105830
UBE3B	100%	100%	100%	100%	99.8%	Kaufman oculocerebrofacial syndrome, 244450
UBE3C	94.8%	94.8%	100%	100%	99.6%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270
UBE4A	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBIAD1	100%	100%	100%	100%	99.7%	Corneal dystrophy, Schnyder type, 121800
UBQLN2	100%	100%	98.3%	90.4%	72.6%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
UBR1	98%	97.9%	100%	100%	99.7%	Johanson-Blizzard syndrome, 243800
UBR2	100%	99.9%	100%	99.8%	99.5%	
UBR5	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with speech delay and behavioral abnormalities, 621372
UBR7	100%	100%	100%	100%	99.4%	Li-Campeau syndrome, 619189
UBTF	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, with brain atrophy, 617672

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
UFC1	100%	100%	100%	99.9%	99.6%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100%	100%	100%	100%	99.8%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
UGCG	100%	100%	100%	99.9%	99.6%	
UGDH	97%	94%	100%	100%	99.7%	Developmental and epileptic encephalopathy 84, 618792
UGGT1	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IICC, 621381
UGP2	91.8%	90.6%	100%	100%	99.8%	Developmental and epileptic encephalopathy 83, 618744

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

UGT1A1	100%	100%	100%	100%	99.6%	Crigler-Najjar syndrome, type I, 218800;[Bilirubin, serum level of, QTL1], 601816;Hyperbilirubinemia, familial transient neonatal, 237900;Crigler-Najjar syndrome, type II, 606785;[Gilbert syndrome], 143500
UMOD	90.8%	89.6%	100%	100%	99.8%	Tubulointerstitial kidney disease, autosomal dominant, 1, 162000
UMPS	90.6%	90.6%	100%	100%	99.6%	Orotic aciduria, 258900
UNC119	100%	100%	100%	100%	99.6%	Cone-rod dystrophy 24, 620342;?Immunodeficiency 13, 615518
UNC13A	100%	100%	100%	99.9%	99%	?Intellectual development disorder with seizures and dysmorphic facies, 621457;Neurodevelopmental disorder with speech delay, movement abnormalities, and seizures, 621456;Neurodevelopmental disorder with hypotonia, epilepsy, and absent speech, 621455
UNC13D	100%	100%	100%	100%	99.5%	Hemophagocytic lymphohistiocytosis, familial, 3, 608898
UNC45A	100%	100%	100%	100%	99.4%	Osteootohepatoenteric syndrome, 619377
UNC45B	96.1%	95.9%	100%	100%	99.7%	?Cataract 43, 616279;Myofibrillar myopathy 11, 619178
UNC79	100%	100%	100%	100%	99.7%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

UNC80	100%	100%	100%	100%	99.7%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UNC93B1	100%	100%	100%	99.7%	98.3%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1}, 610551
UNG	96.5%	96.5%	100%	100%	99.3%	Immunodeficiency with hyper IgM, type 5, 608106
UPB1	94.3%	91.8%	100%	100%	99.5%	Beta-ureidopropionase deficiency, 613161
UPF1	98.7%	96.5%	100%	100%	99.5%	
UPF3B	92.5%	92.5%	99%	90.2%	71.7%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UPK3A	100%	100%	100%	100%	99.7%	
UQCC1	100%	100%	100%	100%	99.7%	
UQCC2	100%	100%	100%	100%	99.9%	Mitochondrial complex III deficiency, nuclear type 7, 615824
UQCC3	100%	100%	100%	100%	99.6%	?Mitochondrial complex III deficiency, nuclear type 9, 616111
UQCR10	100%	100%	100%	100%	99%	
UQCR11	100%	100%	100%	100%	98.8%	
UQCRB	100%	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 3, 615158
UQCRC1	100%	100%	100%	100%	99.5%	Parkinsonism with polyneuropathy, 619279
UQCRC2	74.7%	74.7%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 5, 615160

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

UQCRFS1	100%	100%	100%	100%	99.4%	Mitochondrial complex III deficiency, nuclear type 10, 618775
UQCRH	78.7%	76%	100%	100%	99.8%	?Mitochondrial complex III deficiency, nuclear type 11, 620137
UQCRQ	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 4, 615159
UROC1	96.6%	96.6%	100%	100%	99.6%	?Urocanase deficiency, 276880
UROD	100%	100%	100%	100%	99.6%	Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100
UROS	95.3%	95.3%	100%	100%	99.9%	Porphyria, congenital erythropoietic, 263700
USB1	93.9%	93.2%	100%	100%	99.1%	Poikiloderma with neutropenia, 604173
USH1C	100%	100%	100%	100%	99.6%	Usher syndrome, type 1C, 276904;Deafness, autosomal recessive 18A, 602092
USH1G	100%	100%	100%	100%	99.1%	Usher syndrome, type 1G, 606943
USH2A	100%	100%	100%	100%	99.7%	Usher syndrome, type 2A, 276901;Retinitis pigmentosa 39, 613809
USP14	100%	100%	100%	100%	99.8%	
USP18	100%	100%	100%	100%	99.2%	Pseudo-TORCH syndrome 2, 617397
USP26	100%	100%	99%	91.8%	74.8%	Spermatogenic failure, X-linked, 6, 301101
USP27X	100%	100%	99.3%	92.3%	72.5%	Intellectual developmental disorder, X-linked 105, 300984
USP34	100%	100%	100%	100%	99.9%	
USP45	100%	100%	100%	100%	99.8%	?Leber congenital amaurosis 19, 618513

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

USP48	100%	100%	100%	100%	99.9%	Deafness, autosomal dominant 85, 620227
USP53	100%	100%	100%	100%	99.8%	Cholestasis, progressive familial intrahepatic, 7, with or without hearing loss, 619658
USP7	100%	100%	100%	100%	99.3%	Hao-Fountain syndrome, 616863
USP8	96.3%	96.3%	100%	100%	99.4%	Pituitary adenoma 4, ACTH-secreting, somatic, 219090
USP9X	100%	100%	99.4%	91.8%	75%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
USP9Y	50%	50%	48.9%	40.8%	23.9%	Spermatogenic failure, Y-linked, 2, 415000
UTP4	100%	100%	100%	100%	99.6%	
UVSSA	100%	100%	100%	100%	99.5%	UV-sensitive syndrome 3, 614640
VAC14	96.4%	96.4%	100%	100%	99.7%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

VANGL1	95.2%	91.1%	100%	100%	99.9%	{Neural tube defects, susceptibility to}, 182940;Caudal regression syndrome, 600145
VANGL2	100%	100%	100%	99.9%	99.2%	Neural tube defects, 182940
VAPB	91.4%	91.4%	100%	100%	99.9%	Spinal muscular atrophy, late-onset, Finkel type, 182980;Amyotrophic lateral sclerosis 8, 608627
VARS1	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 20, 615917
VAV1	98.3%	98.3%	100%	100%	99.5%	
VAX1	100%	100%	100%	100%	98.5%	?Microphthalmia, syndromic 11, 614402
VCAN	100%	100%	100%	100%	99.8%	Wagner syndrome 1, 143200
VCL	93.7%	93.6%	100%	100%	99.6%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255
VCP	100%	99.6%	100%	100%	99.8%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954;Charcot-Marie-Tooth disease, type 2Y, 616687;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

VDR	100%	100%	100%	100%	99.1%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100%	100%	100%	100%	99.8%	Lymphatic malformation 4, 615907
VEZF1	86%	86%	100%	99.9%	99.4%	?Cardiomyopathy, dilated, 1OO, 620247
VHL	87.7%	87.7%	100%	99.9%	98.8%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
VIL1	100%	100%	100%	100%	99.1%	
VIM	100%	100%	100%	99.9%	99%	Cataract 30, pulverulent, 116300
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	98.6%	93.5%	100%	100%	99.2%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700
VLDLR	100%	100%	100%	100%	99.8%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VMA12	100%	100%	100%	99.9%	99.2%	Congenital disorder of glycosylation, type IIp, 616829
VMA21	100%	100%	98.5%	86.5%	67.4%	Myopathy, X-linked, with excessive autophagy, 310440
VMA22	100%	98.5%	100%	99.9%	97.9%	Congenital disorder of glycosylation, type IIo, 616828

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

VPS11	100%	100%	100%	100%	99.6%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100%	100%	100%	100%	99.9%	Choreoacanthocytosis, 200150
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS13C	100%	100%	100%	100%	99.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS13D	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100%	100%	100%	100%	99.6%	Dystonia 30, 619291
VPS33A	90.3%	90.2%	100%	100%	99.7%	Mucopolysaccharidosis-plus syndrome, 617303
VPS33B	100%	100%	100%	100%	99.9%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VPS35	100%	100%	100%	100%	99.9%	{Parkinson disease 17}, 614203
VPS35L	93.6%	93%	100%	100%	99.4%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	92.3%	92.3%	100%	99.9%	99.5%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	98.4%	96.8%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS45	88.2%	87.2%	100%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100%	100%	100%	99.9%	98.9%	CIMDAG syndrome, 619273

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

VPS50	96.2%	95.3%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS51	100%	100%	100%	100%	99%	Pontocerebellar hypoplasia, type 13, 618606
VPS53	83.1%	81.5%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99.6%	98%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VSX1	100%	100%	100%	100%	99.6%	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195;Keratoconus 1, 148300
VSX2	100%	100%	100%	99.9%	99.1%	Microphthalmia, isolated 2, 610093;Microphthalmia /coloboma 3, 610092
VTN	100%	100%	100%	100%	99.6%	
VWA1	100%	100%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal recessive 7, 619216
VWA3B	100%	100%	100%	100%	99.7%	?Spinocerebellar ataxia, autosomal recessive 22, 616948
VWA8	100%	100%	100%	100%	99.8%	?Retinitis pigmentosa 97, 620422
VWF	100%	100%	100%	100%	99.5%	von Willebrand disease, type 1, 193400;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 3, 277480

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WAC	94%	94%	100%	100%	99.6%	Desanto-Shinawi syndrome, 616708
WAPL	97.6%	97.6%	100%	100%	99.7%	
WARS1	91.8%	91.8%	100%	100%	99.6%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WARS2	93.8%	93.8%	100%	100%	100%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WAS	99.6%	95.8%	98.5%	87.7%	68.3%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WASF1	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 43, 615817
WASHC5	100%	100%	100%	100%	99.8%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WBP11	100%	100%	100%	100%	99.6%	Vertebral, cardiac, tracheoesophageal, renal, and limb defects, 619227
WBP2	100%	100%	100%	100%	99.3%	Deafness, autosomal recessive 107, 617639
WBP4	100%	100%	100%	100%	99.2%	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852
WDFY3	100%	100%	100%	100%	99.8%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR1	100%	100%	100%	100%	99.4%	Periodic fever, immunodeficiency, and thrombocytopenia syndrome, 150550
WDR11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100%	100%	98%	88%	69.1%	
WDR19	100%	100%	100%	100%	99.8%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WDR26	93.8%	93.8%	100%	100%	99.5%	Skraban-Deardorff syndrome, 617616
WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR36	100%	100%	100%	99.8%	99.4%	Glaucoma 1, open angle, G, 609887
WDR37	90.2%	89.9%	100%	100%	99.8%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	100%	99.9%	99.3%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR44	91.1%	91.1%	99.4%	92.1%	73.3%	
WDR45	98.4%	98%	98.4%	87.9%	68.6%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	89.6%	89.6%	100%	100%	99.6%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR47	100%	100%	100%	100%	99.9%	
WDR5	93.9%	93.9%	100%	100%	99.6%	
WDR62	98.8%	98.8%	100%	100%	99.6%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR72	100%	100%	100%	100%	99.6%	Amelogenesis imperfecta, type IIA3, 613211
WDR73	100%	100%	100%	100%	99.3%	Galloway-Mowat syndrome 1, 251300

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WDR81	100%	100%	100%	100%	99.5%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967
WDR83OS	100%	100%	100%	100%	98.4%	Neurodevelopmental disorder with variable familial hypercholanemia, 621016
WDTC1	100%	100%	100%	100%	99.4%	
WEE2	100%	100%	100%	100%	99.8%	Oocyte/zygote/embryo maturation arrest 5, 617996
WFDC2	100%	100%	100%	100%	98.6%	Bronchiectasis and nasal polyposis, 620984
WFS1	91.5%	90.5%	100%	100%	99.4%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
WHRN	100%	100%	100%	100%	99.4%	Deafness, autosomal recessive 31, 607084;Usher syndrome, type 2D, 611383
WIPF1	100%	100%	100%	100%	99.7%	Wiskott-Aldrich syndrome 2, 614493
WIPI2	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WLS	100%	100%	100%	100%	99.9%	Zaki syndrome, 619648

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WNK1	100%	100%	100%	99.9%	99%	Neuropathy, hereditary sensory and autonomic, type II, 201300;Pseudohypoaldosteronism, type IIC, 614492
WNK3	100%	100%	99.4%	91.4%	73.7%	Prieto syndrome, 309610
WNK4	100%	100%	100%	99.9%	98.7%	Pseudohypoaldosteronism, type IIB, 614491
WNT1	100%	100%	100%	99.9%	99%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WNT10A	79.3%	79.3%	100%	100%	99.7%	Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980
WNT10B	100%	100%	100%	99.7%	98.8%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT2B	96.4%	95.9%	100%	100%	99.8%	Diarrhea 9, 618168
WNT3	100%	100%	100%	99.8%	99.3%	?Tetra-amelia syndrome 1, 273395
WNT4	100%	100%	100%	100%	99.1%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812
WNT5A	100%	100%	100%	99.9%	98.6%	Robinow syndrome, autosomal dominant 1, 180700
WNT6	100%	100%	100%	99.6%	98.3%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WNT7A	100%	100%	100%	100%	99.6%	Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820;?Santos syndrome, 613005
WRAP53	100%	99.9%	100%	99.9%	99.2%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100%	100%	100%	100%	99.7%	Werner syndrome, 277700
WSB2	100%	100%	100%	99.9%	99.1%	Luo-Agrawal neurodevelopmental syndrome, 621552
WT1	100%	100%	100%	99.9%	99.3%	Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070
WWOX	100%	100%	100%	100%	99.7%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XDH	100%	100%	100%	100%	99.6%	Xanthinuria, type I, 278300
XIAP	100%	100%	99.5%	93%	77.1%	Lymphoproliferative syndrome, X-linked, 2, 300635
XIRP2	100%	100%	100%	100%	99.8%	
XIST						X-inactivation, familial skewed, 300087
XK	100%	99.9%	99.4%	92.2%	74%	McLeod syndrome, 300842

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

XKR8	100%	100%	100%	100%	99.6%	
XKRY						
XKRYP7						
XPA	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, group C, 278720
XPNPEP3	100%	100%	100%	99.9%	99.6%	Nephronophthisis-like nephropathy 1, 613159
XPO1	100%	99.9%	100%	100%	99.9%	
XPO5	100%	100%	100%	100%	99.6%	
XPOT	89.4%	89.4%	100%	100%	99.9%	
XPR1	100%	100%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100%	100%	100%	100%	99.2%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
XRCC2	100%	100%	100%	100%	99.9%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
XRCC4	100%	100%	100%	100%	99.9%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100%	100%	100%	99.9%	99.3%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	99.9%	98.8%	100%	99.9%	99.5%	{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

YAP1	92.3%	92.3%	100%	100%	99.6%	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or impaired intellectual development, 120433
YARS1	100%	98.5%	100%	100%	99.5%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YARS2	100%	100%	100%	100%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
YEATS2	100%	100%	100%	100%	99.7%	?Epilepsy, myoclonic, familial adult, 4, 615127
YIF1B	90%	90%	100%	99.9%	99.4%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	100%	100%	100%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	96.7%	96.7%	100%	100%	99.7%	?Optic atrophy 11, 617302
YPEL2	100%	100%	100%	100%	100%	
YRDC	100%	100%	100%	99.9%	98.7%	Galloway-Mowat syndrome 10, 619609
YWHAE	100%	100%	100%	100%	99.9%	
YWHAG	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 56, 617665
YWHAZ	100%	100%	100%	99.9%	99.1%	Popov-Chang syndrome, 618428
YY1	100%	100%	100%	99.6%	96.6%	Gabriele-de Vries syndrome, 617557
YY1AP1	100%	100%	100%	100%	99.9%	Grange syndrome, 602531

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZAP70	100%	100%	100%	100%	99.4%	Immunodeficiency 48, 269840;Autoimmune disease, multisystem, infantile-onset, 2, 617006
ZBTB11	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	87.8%	87.8%	100%	100%	99.3%	Leukemia, acute promyelocytic, PL2F/RARA type
ZBTB17	100%	100%	100%	100%	99.6%	
ZBTB18	93.2%	93.2%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100%	100%	100%	99.9%	99.1%	Primrose syndrome, 259050
ZBTB24	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB42	100%	100%	100%	100%	99.3%	?Lethal congenital contracture syndrome 6, 616248
ZBTB47	99.6%	99.5%	100%	99.9%	98.8%	
ZBTB7A	95.4%	95.4%	100%	99.9%	97.7%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100%	100%	98.3%	88.3%	72.7%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZCCHC8	100%	100%	100%	100%	99.5%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674
ZDHHC9	100%	100%	99%	89.7%	72.4%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799
ZEB1	100%	100%	100%	100%	99.8%	Corneal dystrophy, posterior polymorphous, 3, 609141;Corneal dystrophy, Fuchs endothelial, 6, 613270
ZEB2	100%	100%	100%	99.9%	99.4%	Mowat-Wilson syndrome, 235730
ZFHX2	100%	100%	100%	100%	99.4%	?Marsili syndrome, 147430
ZFHX3	100%	100%	100%	100%	99.4%	Prostate cancer, somatic, 176807;{Atrial fibrillation 8, susceptibility to}, 613055;Spinocerebellar ataxia 4, 600223;Epilepsy, idiopathic generalized 20, 621500
ZFHX4	100%	100%	100%	100%	99.6%	
ZFP36L2	100%	100%	100%	99.7%	98.5%	Oocyte/zygote/embryo maturation arrest 13, 620154
ZFP57	100%	100%	100%	100%	99.5%	Diabetes mellitus, transient neonatal 1, 601410
ZFPM2	100%	100%	100%	100%	99.8%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZFTRAF1	75.9%	72.6%	100%	99.6%	97.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZFX	100%	100%	98.7%	90.4%	73.6%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZFYVE19	100%	100%	100%	100%	99.6%	Cholestasis, progressive familial intrahepatic, 9, 619849
ZFYVE26	100%	100%	100%	100%	99.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100%	100%	100%	99.9%	99.2%	
ZIC1	100%	100%	100%	99.9%	98.6%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	100%	99.9%	99.6%	97%	Holoprosencephaly 5, 609637
ZIC3	100%	100%	98.4%	87.4%	68.6%	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390
ZMIZ1	100%	99.5%	100%	100%	99.3%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZMYM2	100%	100%	100%	100%	99.9%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZMYM3	100%	100%	98.9%	89.9%	71.3%	Intellectual developmental disorder, X-linked 112, 301111
ZMYND10	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 22, 615444
ZMYND11	100%	99.2%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZMYND15	100%	100%	100%	99.9%	99.1%	?Spermatogenic failure 14, 615842
ZMYND8	98%	98%	100%	100%	99.5%	
ZNF124	100%	100%	100%	100%	99.7%	
ZNF141	100%	100%	100%	100%	99.8%	?Polydactyly, postaxial, type A6, 615226
ZNF142	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF143	94.2%	94.2%	100%	100%	99.6%	
ZNF148	94.9%	94.9%	100%	100%	99.8%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99.4%	99.4%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100%	100%	100%	100%	99.5%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF341	100%	100%	100%	100%	99.3%	Hyper-IgE syndrome 3, autosomal recessive, with recurrent infections, 618282
ZNF407	100%	100%	100%	100%	99.8%	SIMHA syndrome, 619557

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZNF408	96.2%	96.2%	100%	100%	99.6%	Retinitis pigmentosa 72, 616469;?Exudative vitreoretinopathy 6, 616468
ZNF41	100%	100%	99.2%	91%	73.3%	
ZNF423	100%	100%	100%	100%	99.4%	Nephronophthisis 14, 614844;Joubert syndrome 19, 614844
ZNF462	100%	100%	100%	100%	99.6%	Weiss-Kruszka syndrome, 618619
ZNF469	100%	100%	100%	99.9%	99.1%	Brittle cornea syndrome 1, 229200
ZNF513	100%	100%	100%	100%	99.4%	?Retinitis pigmentosa 58, 613617
ZNF526	100%	100%	100%	100%	99.3%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF541	100%	100%	100%	100%	99.5%	
ZNF592	100%	100%	100%	100%	99.5%	
ZNF644	100%	100%	100%	100%	99.9%	Myopia 21, autosomal dominant, 614167
ZNF668	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194
ZNF687	100%	100%	100%	100%	99.5%	Paget disease of bone 6, 616833
ZNF699	100%	100%	100%	99.9%	99.3%	DEGCAGS syndrome, 619488
ZNF711	100%	100%	99.3%	91.1%	73.2%	Intellectual developmental disorder, X-linked 97, 300803
ZNF750	100%	100%	100%	100%	99.3%	?Seborrhea-like dermatitis with psoriasiform elements, 610227
ZNF808	100%	100%	100%	100%	99.9%	Pancreatic agenesis 3, 620991

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZNF862	100%	100%	100%	100%	99.6%	?Fibromatosis, gingival 6, 620999
ZNF865	100%	100%	100%	100%	98.9%	
ZNFX1	100%	100%	100%	100%	99.6%	Immunodeficiency 91 and hyperinflammation, 619644
ZNG1A	99.2%	97.9%	96.4%	95.8%	94%	
ZNHIT3	78.8%	76.2%	100%	100%	99.4%	PEHO syndrome, 260565
ZNRF3	98.4%	96.9%	100%	99.9%	99.2%	
ZP1	100%	100%	100%	100%	99.4%	Oocyte/zygote/embryo maturation arrest 1, 615774
ZP2	100%	100%	100%	100%	99.8%	Oocyte/zygote/embryo maturation arrest 6, 618353
ZP3	100%	100%	100%	99.8%	98.8%	Oocyte/zygote/embryo maturation arrest 3, 617712
ZP4	100%	100%	100%	100%	99.4%	
ZPBP	88%	88%	100%	99.9%	99.2%	?Spermatogenic failure 66, 619799
ZPR1	100%	100%	100%	99.9%	99.3%	Growth restriction, hypoplastic kidneys, alopecia, and distinctive facies, 619321
ZRSR2	84.4%	84.4%	98.7%	88.9%	70.2%	Orofaciodigital syndrome XXI, 301132
ZSCAN10	100%	100%	100%	100%	99.2%	Otofacial neurodevelopmental syndrome, 620910
ZSWIM6	97.9%	96.3%	99.2%	98.1%	96.1%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ZSWIM7	91.9%	85.2%	100%	100%	99.6%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors