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

SKIN DISORDERS PANEL[1] DG-5.1.0 (686 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
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
ABCA12	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277
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

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

ABCC6	94.4%	94.4%	100%	100%	99.5%	Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
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
ABHD5	100%	100%	100%	100%	99.9%	Chanarin-Dorfman syndrome, 275630
ACD	100%	100%	100%	99.9%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ACTA2	100%	100%	100%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042

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

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
ACVRL1	91.2%	91.2%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	95.1%	93.2%	100%	100%	99.8%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
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
ADAMTS10	100%	100%	100%	100%	99.6%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100%	100%	100%	100%	99.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS2	97.9%	97.9%	100%	99.9%	99.5%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410

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

ADAMTS3	100%	100%	100%	100%	99.8%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	100%	100%	100%	99.9%	99.4%	Geleophysic dysplasia 1, 231050
ADAR	100%	100%	100%	100%	99.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
AGA	91.6%	91.6%	100%	100%	99.5%	Aspartylglucosaminuria, 208400
AGPAT2	100%	99.9%	100%	100%	98.9%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
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
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

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

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
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200
ALDOB	100%	100%	100%	99.9%	99.8%	Fructose intolerance, hereditary, 229600
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
ALPL	100%	100%	100%	100%	99.3%	Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALX4	100%	100%	100%	100%	99.4%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMELX	100%	100%	98.3%	89.3%	72.6%	Amelogenesis imperfecta, type 1E, 301200
ANGPT2	100%	100%	100%	100%	99.7%	Lymphatic malformation 10, 619369
ANKRD11	100%	100%	100%	100%	99.2%	KBG syndrome, 148050

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
AP1B1	93.7%	91.9%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S3	90.6%	90.6%	100%	100%	99.9%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 2, 608233
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
APCDD1	100%	100%	100%	100%	99.5%	Hypotrichosis 1, 605389
AQP5	100%	100%	100%	100%	99.5%	Palmoplantar keratoderma, Bothnian type, 600231

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

ARHGAP31	100%	100%	100%	100%	99.6%	Adams-Oliver syndrome 1, 100300
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
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
ASPRV1	100%	100%	100%	100%	99.2%	Ichthyosis, lamellar, autosomal dominant, 146750
ASXL1	100%	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL3	100%	100%	100%	100%	99.6%	Bainbridge-Ropers syndrome, 615485
ATIC	100%	100%	100%	100%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	100%	100%	100%	100%	99.6%	Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236
ATP2C1	96.2%	96.2%	100%	100%	99.8%	Hailey-Hailey disease, 169600
ATP6V0A2	100%	100%	100%	100%	99.6%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200

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

ATP7A	95%	94.8%	99.4%	92.2%	74.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATR	100%	100%	99.9%	99.7%	99.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100%	100%	100%	100%	99.2%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
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
B4GALT7	86.8%	86.8%	100%	100%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
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
BCOR	100%	100%	98.9%	89.4%	69.8%	Microphthalmia, syndromic 2, 300166

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

BCS1L	100%	100%	100%	100%	99.6%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BLM	94.7%	94.6%	100%	100%	99.9%	Bloom syndrome, 210900
BLOC1S3	100%	100%	100%	100%	98.5%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 9, 614171
BMS1	100%	100%	100%	100%	99.8%	?Aplasia cutis congenita, nonsyndromic, 107600
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
BRIP1	96%	96%	100%	100%	99.8%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480

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

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
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260
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
C1QC	99.7%	98%	100%	100%	98.7%	C1q deficiency 3, 620322
C2CD3	96%	96%	100%	100%	99.5%	Orofaciodigital syndrome XIV, 615948
CA2	100%	100%	100%	100%	99.8%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	100%	100%	100%	99.9%	98.6%	
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
CARD9	100%	100%	100%	100%	99.6%	Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	100%	100%	100%	100%	99.2%	Immunodeficiency 58, 618131

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

CARS1	100%	100%	100%	100%	99.6%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CASP14	100%	100%	100%	100%	99.6%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	100%	100%	100%	100%	99.8%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
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
CAVIN1	100%	100%	100%	99.6%	97.4%	Lipodystrophy, congenital generalized, type 4, 613327
CAVIN2	100%	100%	100%	100%	99.7%	
CBL	100%	100%	100%	100%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CCBE1	100%	100%	100%	100%	99.7%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510

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

CD151	100%	100%	100%	100%	99.7%	[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CDAN1	100%	100%	100%	100%	98.9%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	100%	100%	100%	100%	99.6%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDSN	100%	100%	100%	100%	99.5%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300
CELSR1	100%	100%	100%	100%	99.2%	Lymphatic malformation 9, 619319;Yellow nail syndrome, 153300
CERS3	100%	100%	100%	100%	99.8%	Ichthyosis, congenital, autosomal recessive 9, 615023
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}
CHKB	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776

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

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
CIB1	100%	100%	100%	100%	99.6%	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267
CKAP2L	100%	100%	100%	100%	99.7%	Filippi syndrome, 272440
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
CNNM4	100%	100%	100%	100%	99.4%	Jalili syndrome, 217080
COG6	100%	100%	100%	100%	99.9%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
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.

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
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
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

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
COX4I2	100%	99.1%	100%	100%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	100%	100%	98.7%	86.9%	70%	Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	93.4%	93.4%	100%	99.9%	99%	Coproporphyria, 121300;Harderoporphyria, 618892
CRIPT	100%	100%	100%	100%	99.9%	Rothmund-Thomson syndrome, type 3, 615789
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
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

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
CTSZ	81.3%	73.5%	100%	99.9%	99.6%	
CXCR4	99%	99%	100%	100%	99.5%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
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
CYP26C1	100%	100%	100%	100%	99.4%	Focal facial dermal dysplasia 4, 614974
CYP4F22	100%	100%	100%	100%	99.1%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	97.1%	97.1%	100%	100%	99.9%	Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DDB2	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740

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

DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DKC1	100%	100%	99%	89.5%	70.6%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLX3	100%	100%	100%	99.9%	99.4%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX5	100%	100%	100%	100%	99.2%	Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	100%	100%	100%	99.9%	99.3%	Adams-Oliver syndrome 2, 614219
DOCK8	98.6%	98.6%	100%	100%	99.7%	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DOLK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type Im, 610768
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

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

DSG1	100%	100%	100%	100%	99.8%	Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
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
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

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

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
DTNBP1	96.4%	96.4%	100%	100%	99.7%	Hermansky-Pudlak syndrome 7, 614076
DUSP1	100%	100%	100%	100%	99.6%	
DUSP6	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	100%	100%	98.8%	88.4%	69.5%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECM1	100%	100%	100%	100%	99.3%	Urbach-Wiethe disease, 247100
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

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

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
EDN3	100%	100%	100%	99.9%	99.4%	Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
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
EFEMP2	99.6%	95.2%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	100%	100%	98.8%	87.7%	69.3%	Craniofrontonasal dysplasia, 304110
EIF2AK3	100%	100%	100%	100%	99.7%	Wolcott-Rallison syndrome, 226980
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

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

ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
EMP2	100%	100%	100%	100%	98.7%	Nephrotic syndrome, type 10, 615861
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
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
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHB4	97.7%	95.6%	100%	100%	99.3%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
EPHX3	100%	100%	100%	100%	99.5%	
EPS8L3	100%	99.9%	100%	100%	99.3%	?Hypotrichosis 5, 612841

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

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
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

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

ERCC8	95.2%	95.2%	100%	99.9%	98.8%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERG	100%	100%	100%	100%	99.4%	Lymphatic malformation 14, 620602
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
EXPH5	100%	100%	100%	100%	99.8%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
F13A1	100%	100%	100%	100%	99.7%	Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050
FAM111B	100%	100%	100%	100%	99.4%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	100%	99.9%	100%	99.8%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	99.4%	97.2%	100%	100%	99.2%	Raine syndrome, 259775
FAM83G	100%	100%	100%	100%	99.6%	
FAM83H	100%	100%	100%	100%	99.3%	

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

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
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
FAT4	100%	100%	100%	99.9%	99.5%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006

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

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
FDFT1	100%	98.7%	100%	100%	99.5%	Squalene synthase deficiency, 618156
FDPS	100%	100%	100%	100%	99.3%	Porokeratosis 9, multiple types, 616631
FECH	100%	100%	100%	100%	99.8%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	100%	100%	100%	100%	99.7%	Kindler syndrome, 173650
FGF10	100%	100%	100%	100%	99.9%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
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
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

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

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
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
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
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
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

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

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
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
FNIP1	100%	100%	100%	99.9%	99.7%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
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
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
FOXP3	100%	100%	99.1%	88.7%	71.1%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790

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

FREM1	100%	100%	100%	100%	99.6%	Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FUCA1	100%	100%	100%	100%	99.5%	Fucosidosis, 230000
FZD6	99.9%	99.1%	100%	100%	99.8%	Nail disorder, nonsyndromic congenital, 1, 161050
GALNS	100%	100%	100%	100%	99%	Mucopolysaccharidosis IVA, 253000
GALNT3	100%	100%	100%	100%	99.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	100%	100%	100%	99.9%	99.5%	Giant axonal neuropathy-1, 256850
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
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

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

GDF2	100%	100%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
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
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
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

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

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
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
GLA	91.4%	91.4%	99.3%	91.7%	72.9%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500

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

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
GLMN	96.2%	96.2%	100%	100%	99.9%	Glomuvenous malformations, 138000
GMPPA	100%	100%	100%	100%	98.8%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510
GNA11	100%	100%	100%	100%	99.5%	Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361
GNA14	100%	100%	100%	100%	99.9%	
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

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

GORAB	100%	100%	100%	100%	99.5%	Geroderma osteodysplasticum, 231070
GPNMB	95.1%	95.1%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 3, 617920
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
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
GSN	92.1%	90%	100%	99.8%	98.9%	Amyloidosis, Finnish type, 105120
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
HCCS	100%	100%	99.3%	91.9%	75.5%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	94.6%	94.6%	99.2%	90.3%	71.3%	Cornelia de Lange syndrome 5, 300882

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

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
HLCS	100%	100%	100%	100%	99.2%	Holocarboxylase synthetase deficiency, 253270
HMBS	100%	100%	100%	100%	99.9%	Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, 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
HMGB3	100%	100%	99.1%	89.3%	70.4%	?Microphthalmia, syndromic 13, 300915
HOXC13	100%	100%	100%	100%	99.4%	Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	100%	100%	100%	100%	99.9%	?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100
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

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

HPS5	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	100%	99.4%	Hermansky-Pudlak syndrome 6, 614075
HR	100%	100%	100%	100%	99.5%	Atrichia with papular lesions, 209500;Alopecia universalis, 203655
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
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
HYAL1	100%	100%	100%	99.9%	98.9%	Mucopolysaccharidosis type IX, 601492
IDUA	96%	96%	100%	99.8%	98.7%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IFT122	100%	100%	100%	100%	99.6%	Cranioectodermal dysplasia 1, 218330

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

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
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
IL17RA	100%	100%	100%	100%	99.5%	Immunodeficiency 51, 613953
IL17RD	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
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
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.

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
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
ISG15	100%	100%	100%	100%	99.6%	Immunodeficiency 38, 616126
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
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
JUP	100%	100%	100%	100%	99.5%	Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528

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

KANK2	100%	100%	100%	100%	99.6%	Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KAT6B	100%	100%	100%	100%	99.5%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KCNH1	98.6%	98.6%	100%	100%	99.8%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNK9	100%	100%	100%	100%	99.8%	Birk-Barel syndrome, 612292
KCTD1	100%	100%	100%	99.8%	98.3%	Scalp-ear-nipple syndrome, 181270;Dental radicular dysplasia, 621434
KDF1	100%	100%	100%	100%	98.8%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	91.6%	91.6%	100%	100%	98.4%	Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	100%	100%	100%	100%	99.8%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
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

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

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
KLF4	100%	100%	100%	100%	99.3%	
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
KLK11	100%	100%	100%	100%	98.8%	Ichthyosis with erythrokeratoderma, 620507
KLK4	100%	100%	100%	99.9%	99.2%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	100%	100%	100%	100%	99.2%	Cowden syndrome 4, 615107
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920

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
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

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

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
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-Franceschetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900
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
KRT2	100%	100%	100%	100%	99.7%	Ichthyosis bullosa of Siemens, 146800
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
KRT81	100%	100%	100%	100%	98.6%	Monilethrix 2, 621169
KRT82	100%	100%	100%	100%	99.5%	

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

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
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
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
LAMC2	100%	100%	100%	99.9%	99.5%	Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMTOR2	96.6%	85.3%	100%	100%	99.3%	?Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	100%	100%	100%	100%	99.6%	Glycogen storage disease XI, 612933
LDLRAP1	93.2%	90.2%	100%	100%	99.2%	Hypercholesterolemia, familial, 4, 603813

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

LEMD3	100%	100%	100%	99.9%	99%	Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
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
LMBRD1	86.4%	86.4%	100%	99.9%	99.6%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
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
LMX1B	100%	100%	100%	100%	99%	Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LONP1	99.3%	97%	100%	100%	99.5%	CODAS syndrome, 600373

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

LORICRIN	100%	100%	100%	99.9%	97.4%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	100%	100%	100%	100%	99.9%	Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	99.3%	99.2%	100%	99.9%	99.4%	Majeed syndrome, 609628
LRMDA	97.8%	97.8%	100%	100%	99.4%	Albinism, oculocutaneous, type VII, 615179
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
LSS	100%	100%	100%	100%	99.8%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840
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
LTV1	100%	100%	100%	100%	99.9%	Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199
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

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

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
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
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
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
MDFIC	100%	100%	100%	100%	99.8%	Lymphatic malformation 12, 620014
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

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

MEFV	96.1%	96.1%	100%	100%	99.6%	Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MGP	100%	100%	100%	100%	99.7%	Keutel syndrome, 245150
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
MLH1	100%	100%	100%	100%	99.7%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLPH	95.4%	95.4%	100%	100%	99.3%	Griscelli syndrome, type 3, 609227
MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP1	100%	100%	100%	100%	99.8%	
MMP14	94.9%	94.9%	100%	100%	99.5%	Winchester syndrome, 277950
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
MPDU1	100%	100%	100%	100%	98.8%	Congenital disorder of glycosylation, type If, 609180

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

MPLKIP	100%	100%	100%	100%	99%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11	97.4%	97.4%	100%	100%	99.9%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	90.6%	90.6%	100%	100%	99.6%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
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
MTOR	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
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
MYH8	100%	100%	100%	100%	99.6%	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	99%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450
NAA10	100%	100%	99%	89%	68.4%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855

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

NAGA	90%	88%	100%	100%	99.5%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241
NBAS	100%	100%	100%	100%	99.7%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483
NCF1	99.7%	96.1%	100%	99.7%	97.8%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCSTN	100%	100%	100%	100%	99.5%	Acne inversa, familial, 1, 142690
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
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
NEK11	87.7%	87.7%	100%	100%	99.7%	
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

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

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
NFKBIA	100%	100%	100%	100%	99.3%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal recessive 2, 613987
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
NKPD1	100%	100%	100%	99.9%	98.5%	
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

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

NLRP3	100%	100%	100%	100%	99.6%	CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendotheliti fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900
NME1	100%	100%	100%	100%	99.8%	
NOD2	100%	100%	100%	100%	99.7%	Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600
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
NOTCH1	99.1%	99%	100%	99.9%	99.4%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730

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

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
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSDHL	95.9%	95.9%	99.3%	93.1%	75.4%	CK syndrome, 300831;CHILD syndrome, 308050
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
ODAM	100%	100%	100%	100%	99.5%	
ODAPH	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA4, 614832

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
OSMR	100%	100%	100%	100%	99.8%	Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	100%	100%	100%	100%	99.6%	Uncombable hair syndrome, 191480
PAH	94.9%	93.7%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
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
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
PAX9	100%	100%	100%	100%	99.4%	Tooth agenesis, selective, 3, 604625
PCNA	100%	100%	100%	100%	99.8%	?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	99.7%	94.7%	100%	100%	99.8%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483

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

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
PEPD	94%	94%	100%	100%	99.6%	Prolidase deficiency, 170100
PER3	100%	100%	100%	100%	99.5%	?Advanced sleep phase syndrome, familial, 3, 616882
PERP	100%	100%	100%	100%	99.4%	Erythrokeratodermia variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHEX	100%	100%	99%	92.2%	75%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	100%	100%	100%	100%	99.6%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500

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

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
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
PIGL	100%	100%	100%	100%	99.7%	CHIME syndrome, 280000
PIGN	95.9%	94.4%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100%	100%	100%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300

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
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

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

PITX2	100%	100%	100%	100%	99.1%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKP1	100%	100%	100%	100%	99.6%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	100%	100%	100%	100%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	96.3%	96.3%	100%	100%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
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
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.

PLOD1	96.5%	96.5%	100%	100%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	98.5%	96.8%	100%	99.8%	98.9%	BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PMS2	93.4%	93.4%	99.8%	99.6%	98.9%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PMVK	71.2%	71.2%	100%	100%	99.5%	Porokeratosis 1, multiple types, 175800
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
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
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

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

POLA1	100%	100%	99.3%	91.7%	73.3%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030
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
POLH	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, variant type, 278750
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
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
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

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

POMC	100%	100%	100%	100%	99.7%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMP	83.2%	83.2%	100%	100%	99.6%	Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	100%	100%	98.5%	89.4%	70.4%	Focal dermal hypoplasia, 305600
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
PPOX	99.9%	97.6%	100%	100%	99.3%	Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PQBP1	100%	100%	98.6%	86.7%	67.3%	Renpenning syndrome, 309500
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

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
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
PSENEN	100%	100%	100%	100%	99.8%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	100%	100%	100%	99.9%	99.2%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
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

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

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
PTDSS1	93%	93%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
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
PTHLH	100%	100%	100%	100%	99.8%	Brachydactyly, type E2, 613382
PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	100%	100%	100%	100%	99.5%	Choanal atresia and lymphedema, 613611
PTPRF	100%	100%	100%	100%	99.4%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
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.

RAB23	100%	100%	100%	100%	99.4%	Carpenter syndrome 1, 201000
RAB27A	100%	100%	100%	100%	99.6%	Griscelli syndrome, type 2, 607624
RAD21	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD50	100%	100%	100%	100%	99.7%	Nijmegen breakage syndrome-like disorder, 613078
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.

RASA1	100%	100%	100%	100%	99.8%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RBBP8	100%	100%	100%	100%	99.8%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBM28	100%	100%	100%	100%	99.7%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
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
RECQL4	100%	100%	100%	100%	99.4%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RHBDF2	100%	100%	100%	99.9%	98.5%	Tylosis with esophageal cancer, 148500
RHOA	81.7%	81.7%	100%	100%	99.6%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RIN2	98.3%	98.3%	100%	100%	99.2%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075

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

RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355
RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
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
RNF113A	100%	100%	99.3%	87.4%	70.4%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROGDI	100%	100%	100%	99.9%	98.8%	Kohlschutter-Tonz syndrome, 226750
RPL10A	100%	100%	100%	100%	99.2%	
RPL21	100%	100%	100%	100%	99.8%	Hypotrichosis 12, 615885
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

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

RSPO4	100%	100%	100%	100%	99.4%	Anonychia congenita, 206800
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
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
S100A3	100%	100%	100%	100%	99.4%	
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMHD1	92%	89.8%	100%	100%	99.5%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
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
SAT1	100%	100%	98.9%	89.4%	69.8%	

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

SATB2	100%	100%	100%	99.9%	99.5%	Glass syndrome, 612313
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
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
SDR9C7	100%	100%	100%	100%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	100%	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SERPINA12	100%	100%	100%	100%	99.8%	
SERPINA3	100%	100%	100%	100%	99.9%	Alpha-1-antichymotrypsin deficiency;Cerebrovascular disease, occlusive
SERPINB7	100%	100%	100%	100%	99.7%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100%	100%	100%	100%	99.6%	Peeling skin syndrome 5, 617115
SERPING1	100%	100%	100%	100%	99.6%	Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790

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

SERPINH1	100%	100%	100%	100%	99.6%	{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SHOC2	100%	100%	100%	100%	99.9%	Noonan syndrome-like with loose anagen hair 1, 607721
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
SLC17A9	99.9%	97.7%	100%	100%	99.5%	Porokeratosis 8, disseminated superficial actinic type, 616063
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
SLC24A5	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750

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

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
SLC27A4	100%	100%	100%	100%	99.6%	Ichthyosis prematurity syndrome, 608649
SLC29A3	100%	100%	100%	100%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	94.7%	94.7%	100%	100%	99.8%	Arterial tortuosity syndrome, 208050
SLC39A13	99.8%	97.5%	100%	100%	99.5%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	100%	100%	100%	100%	99.5%	Acrodermatitis enteropathica, 201100
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
SLC4A4	95%	95%	100%	100%	99.9%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC7A7	100%	100%	100%	100%	98.8%	Lysinuric protein intolerance, 222700

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

SLCO2A1	100%	100%	100%	100%	99.3%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLURP1	100%	100%	100%	100%	99.2%	Meleda disease, 248300
SLX4	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group P, 613951
SMAD3	100%	100%	100%	100%	99.4%	Loeys-Dietz syndrome 3, 613795
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
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

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

SMO	100%	100%	100%	100%	99.7%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	100%	100%	100%	100%	99.6%	Dentin dysplasia, type IA, 125400
SNAI2	100%	100%	100%	100%	99.7%	
SNAP29	100%	100%	100%	99.7%	99.3%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	100%	100%	100%	100%	99.9%	Hypotrichosis 11, 615059
SNX10	89.3%	89.3%	100%	100%	99.6%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
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
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

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

SP7	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type XII, 613849
SPINK5	100%	100%	100%	100%	99.8%	Netherton syndrome, 256500
SPINT2	100%	100%	100%	100%	99.2%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	97.6%	97.6%	100%	100%	100%	Legius syndrome, 611431
SPRY4	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
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
ST14	100%	100%	100%	100%	99.5%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	98.4%	98.4%	100%	100%	99.6%	Salt and pepper developmental regression syndrome, 609056
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261
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.

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
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
STK4	100%	100%	100%	100%	100%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STS	97.1%	97.1%	98.9%	90.5%	73.5%	Ichthyosis, X-linked, 308100
SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SULT2B1	100%	100%	100%	100%	98.7%	Ichthyosis, congenital, autosomal recessive 14, 617571

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

SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200
TAB3	100%	100%	99.4%	91.3%	73.6%	
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
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
TARS1	100%	100%	100%	100%	99.9%	Trichothiodystrophy 7, nonphotosensitive, 618546
TAT	100%	100%	100%	100%	99.8%	Tyrosinemia, type II, 276600
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
TBX3	100%	100%	100%	100%	99.2%	Ulnar-mammary syndrome, 181450
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

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

TEK	100%	100%	100%	100%	99.9%	Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272
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
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
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
TGFB2	100%	100%	100%	99.8%	99.4%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631

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

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
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
TINF2	100%	100%	100%	100%	99.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TMC6	94.9%	94.4%	100%	99.9%	99.1%	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400
TMC8	100%	100%	100%	100%	99.4%	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TMEM165	88.9%	88.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
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.

TNFRSF1A	100%	100%	100%	100%	99.6%	{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680
TNFSF11	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	100%	100%	100%	99.9%	98.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
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
TPCN2	100%	100%	100%	100%	99.5%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
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

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

TRIM32	100%	100%	100%	100%	99.6%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	98.3%	97.6%	100%	100%	99.7%	Mulibrey nanism, 253250
TRPM4	100%	100%	100%	100%	99.4%	Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531
TRPS1	100%	100%	100%	100%	99.8%	Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350
TRPV3	96.6%	96.6%	100%	100%	99.3%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594
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
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

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

TTI2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 39, 615541
TUFT1	100%	100%	100%	100%	99.4%	Woolly hair-skin fragility syndrome, 620415
TWIST2	100%	100%	100%	99.9%	96.6%	Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260
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
TYRP1	100%	100%	100%	100%	99.7%	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
UBA2	100%	100%	100%	100%	99.6%	ACCES syndrome, 619959
UBE2A	95.6%	91.8%	99%	90.8%	72.2%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBR1	98%	97.9%	100%	100%	99.7%	Johanson-Blizzard syndrome, 243800
UGCG	100%	100%	100%	99.9%	99.6%	

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

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
UVSSA	100%	100%	100%	100%	99.5%	UV-sensitive syndrome 3, 614640
VANGL2	100%	100%	100%	99.9%	99.2%	Neural tube defects, 182940
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
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
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
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

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

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
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
WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR72	100%	100%	100%	100%	99.6%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	100%	100%	100%	100%	99.7%	Wiskott-Aldrich syndrome 2, 614493
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
WNT5A	100%	100%	100%	99.9%	98.6%	Robinow syndrome, autosomal dominant 1, 180700

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
XPA	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, group C, 278720
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
YWHAZ	100%	100%	100%	99.9%	99.1%	Popov-Chang syndrome, 618428
ZBTB20	100%	100%	100%	99.9%	99.1%	Primrose syndrome, 259050
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZNF469	100%	100%	100%	99.9%	99.1%	Brittle cornea syndrome 1, 229200
ZNF592	100%	100%	100%	100%	99.5%	
ZNF750	100%	100%	100%	100%	99.3%	?Seborrhea-like dermatitis with psoriasiform elements, 610227

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

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