WES INTELLECTUAL DISABILITY DG 3.8.1

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100.0%	100.0%	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100.0%	100.0%	100.0%	99.5%	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
AASS	100.0%	100.0%	100.0%	98.9%	Hyperlysinemia, 238700
ABAT	100.0%	100.0%	100.0%	99.6%	GABA-transaminase deficiency, 613163
ABCA2	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808
ABCC8	100.0%	100.0%	100.0%	99.7%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450

ABCC9	100.0%	100.0%	100.0%	99.4%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABCD1	100.0%	99.6%	99.5%	83.3%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABCD4	100.0%	100.0%	100.0%	99.6%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD16A	100.0%	100.0%	100.0%	99.5%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100.0%	100.0%	100.0%	99.7%	Chanarin-Dorfman syndrome, 275630
ACAD9	100.0%	100.0%	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	100.0%	100.0%	100.0%	100.0%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470
ACADSB	100.0%	100.0%	100.0%	99.5%	2-methylbutyrylglycinuria, 610006
ACAT1	100.0%	100.0%	99.8%	97.2%	Alpha-methylacetoacetic aciduria, 203750
ACER3	100.0%	100.0%	100.0%	99.4%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	100.0%	100.0%	100.0%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	100.0%	100.0%	100.0%	99.3%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	100.0%	100.0%	100.0%	99.5%	Combined malonic and methylmalonic aciduria, 614265

ACSL4	100.0%	100.0%	98.5%	76.3%	Intellectual developmental disorder, X-linked 63, 300387
ACTB	100.0%	100.0%	100.0%	99.9%	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, 620479
ACTG1	100.0%	100.0%	100.0%	99.8%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACTL6A	100.0%	100.0%	100.0%	98.9%	
ACTL6B	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACVR1	100.0%	99.9%	100.0%	99.7%	Fibrodysplasia ossificans progressiva, 135100
ACY1	100.0%	100.0%	100.0%	99.8%	Aminoacylase 1 deficiency, 609924
ADAM22	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 61, 617933
ADAR	100.0%	100.0%	100.0%	99.1%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADARB1	95.0%	94.8%	100.0%	99.8%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100.0%	100.0%	100.0%	100.0%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286

ADCY5	100.0%	99.9%	100.0%	99.6%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADD1	100.0%	100.0%	100.0%	99.8%	{Hypertension, essential, salt-sensitive}, 145500
ADD3	100.0%	100.0%	100.0%	99.3%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100.0%	100.0%	100.0%	99.8%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854
ADK	90.9%	90.9%	100.0%	99.5%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	100.0%	100.0%	100.0%	99.4%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100.0%	100.0%	100.0%	99.8%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	100.0%	100.0%	100.0%	99.4%	Adenylosuccinase deficiency, 103050
AFF2	100.0%	99.8%	98.3%	72.3%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100.0%	100.0%	100.0%	99.4%	KINSSHIP syndrome, 619297
AFF4	100.0%	100.0%	100.0%	99.2%	CHOPS syndrome, 616368
AFG3L2	100.0%	100.0%	100.0%	99.1%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246

AGA	100.0%	100.0%	100.0%	99.6%	Aspartylglucosaminuria, 208400
AGAP1	100.0%	100.0%	100.0%	97.8%	
AGMO	100.0%	100.0%	100.0%	99.2%	
AGO1	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292
AGO2	100.0%	99.9%	100.0%	99.6%	Lessel-Kreienkamp syndrome, 619149
AGTPBP1	100.0%	100.0%	100.0%	99.1%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AHCY	100.0%	100.0%	100.0%	99.9%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100.0%	100.0%	100.0%	99.4%	Xia-Gibbs syndrome, 615829
AHI1	100.0%	100.0%	100.0%	99.1%	Joubert syndrome 3, 608629
AHSG	100.0%	100.0%	100.0%	99.4%	?Alopecia-intellectual disability syndrome 1, 203650
AIFM1	100.0%	99.9%	98.3%	73.3%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AIMP1	100.0%	100.0%	100.0%	98.9%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100.0%	100.0%	100.0%	99.6%	Leukodystrophy, hypomyelinating, 17, 618006
AKT3	100.0%	99.9%	100.0%	99.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937

ALDH18A1	100.0%	100.0%	100.0%	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%	100.0%	99.5%	Sjogren-Larsson syndrome, 270200
ALDH4A1	100.0%	100.0%	100.0%	99.6%	Hyperprolinemia, type II, 239510
ALDH5A1	100.0%	100.0%	100.0%	99.6%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100.0%	100.0%	100.0%	99.6%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALG1	100.0%	100.0%	100.0%	99.9%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96.0%	96.0%	100.0%	99.5%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100.0%	100.0%	100.0%	99.9%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	99.7%	99.0%	97.8%	72.7%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100.0%	100.0%	100.0%	99.1%	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	100.0%	100.0%	100.0%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228
ALG3	100.0%	100.0%	100.0%	99.7%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100.0%	100.0%	100.0%	98.8%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	96.1%	96.1%	100.0%	99.1%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100.0%	100.0%	100.0%	99.3%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALKBH8	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100.0%	100.0%	100.0%	99.2%	Alstrom syndrome, 203800
ALX3	100.0%	100.0%	100.0%	99.3%	Frontonasal dysplasia 1, 136760
ALX4	100.0%	100.0%	100.0%	99.3%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMER1	100.0%	100.0%	99.3%	79.1%	Osteopathia striata with cranial sclerosis, 300373
AMFR	100.0%	100.0%	100.0%	99.7%	Spastic paraplegia 89, autosomal recessive, 620379
AMMECR1	100.0%	99.8%	98.5%	70.6%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMOTL1	100.0%	100.0%	100.0%	99.6%	

AMPD2	100.0%	100.0%	100.0%	99.4%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
AMT	100.0%	100.0%	100.0%	99.8%	Glycine encephalopathy 2, 620398
ANK2	100.0%	100.0%	100.0%	99.4%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	100.0%	99.9%	100.0%	99.2%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKH	100.0%	100.0%	100.0%	99.9%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKLE2	100.0%	100.0%	100.0%	98.1%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	100.0%	100.0%	100.0%	98.8%	KBG syndrome, 148050
ANKRD17	100.0%	100.0%	100.0%	99.3%	Chopra-Amiel-Gordon syndrome, 619504
ANKS1B	100.0%	100.0%	100.0%	99.3%	
ANO10	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANTXR1	100.0%	99.8%	99.4%	94.8%	GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
AP1G1	100.0%	100.0%	100.0%	99.5%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100.0%	100.0%	100.0%	99.5%	MEDNIK syndrome, 609313
AP1S2	100.0%	100.0%	98.3%	72.9%	Pettigrew syndrome, 304340
AP2M1	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder 60 with seizures, 618587

AP2S1	100.0%	100.0%	100.0%	96.1%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100.0%	100.0%	100.0%	99.7%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100.0%	100.0%	100.0%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100.0%	100.0%	100.0%	99.7%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100.0%	100.0%	100.0%	99.4%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100.0%	100.0%	100.0%	99.7%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	87.4%	87.4%	100.0%	99.2%	Spastic paraplegia 52, autosomal recessive, 614067
APC2	100.0%	100.0%	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
APTX	100.0%	100.0%	100.0%	99.5%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	100.0%	100.0%	100.0%	99.6%	Short stature-micrognathia syndrome, 617164
ARF1	100.0%	100.0%	100.0%	100.0%	Periventricular nodular heterotopia 8, 618185
ARF3	100.0%	100.0%	100.0%	99.2%	
ARFGEF1	100.0%	100.0%	100.0%	98.9%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964

ARFGEF2	100.0%	100.0%	100.0%	99.5%	Periventricular heterotopia with microcephaly, 608097
ARG1	93.0%	93.0%	100.0%	99.7%	Argininemia, 207800
ARHGAP31	100.0%	100.0%	100.0%	99.3%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100.0%	100.0%	100.0%	99.4%	
ARHGEF6	100.0%	100.0%	98.3%	72.0%	
ARHGEF9	96.7%	95.8%	98.9%	75.3%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100.0%	100.0%	100.0%	98.1%	Coffin-Siris syndrome 2, 614607
ARID1B	98.6%	98.3%	99.9%	95.8%	Coffin-Siris syndrome 1, 135900
ARID2	100.0%	100.0%	100.0%	99.0%	Coffin-Siris syndrome 6, 617808
ARL13B	100.0%	100.0%	100.0%	98.5%	Joubert syndrome 8, 612291
ARL6	100.0%	100.0%	100.0%	98.1%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARMC9	100.0%	100.0%	100.0%	99.6%	Joubert syndrome 30, 617622
ARPC4	100.0%	100.0%	100.0%	99.6%	Developmental delay, language impairment, and ocular abnormalities, 620141
ARSA	100.0%	100.0%	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARSL	100.0%	100.0%	98.4%	72.7%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 38, 617020

ARX	99.0%	96.7%	93.9%	63.3%	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	100.0%	100.0%	100.0%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASH1L	98.6%	98.6%	100.0%	99.2%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASL	100.0%	100.0%	100.0%	99.9%	Argininosuccinic aciduria, 207900
ASNS	100.0%	100.0%	100.0%	99.4%	Asparagine synthetase deficiency, 615574
ASPA	100.0%	100.0%	100.0%	99.0%	Canavan disease, 271900
ASPM	100.0%	99.8%	100.0%	99.2%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	100.0%	100.0%	100.0%	99.9%	Citrullinemia, 215700
ASXL1	100.0%	100.0%	100.0%	99.6%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL2	100.0%	100.0%	100.0%	99.2%	Shashi-Pena syndrome, 617190
ASXL3	100.0%	100.0%	100.0%	98.8%	Bainbridge-Ropers syndrome, 615485
ATAD1	100.0%	99.7%	100.0%	98.9%	Hyperekplexia 4, 618011
ATAD3A	100.0%	100.0%	100.0%	99.0%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATG4D	100.0%	100.0%	100.0%	99.3%	

ATG7	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100.0%	100.0%	100.0%	99.2%	AICA-ribosiduria due to ATIC deficiency, 608688
ATL1	100.0%	100.0%	100.0%	99.0%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATN1	100.0%	100.0%	100.0%	98.9%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP13A2	100.0%	100.0%	100.0%	99.8%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A1	100.0%	100.0%	100.0%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100.0%	100.0%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.5%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2A2	100.0%	100.0%	100.0%	99.5%	Acrokeratosis verruciformis, 101900;Darier disease, 124200

ATP2B1	100.0%	100.0%	100.0%	98.9%	Intellectual developmental disorder, autosomal dominant 66, 619910
ATP5F1A	100.0%	100.0%	100.0%	99.6%	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
ATP6AP1	100.0%	99.7%	99.2%	77.2%	Immunodeficiency 47, 300972
ATP6AP2	100.0%	100.0%	98.9%	74.3%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0A2	100.0%	100.0%	100.0%	98.9%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0C	100.0%	100.0%	100.0%	100.0%	Epilepsy, early-onset, 3, with or without developmental delay, 620465
ATP6V1A	100.0%	100.0%	100.0%	99.3%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012

ATP6V1B2	100.0%	100.0%	100.0%	99.4%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP7A	100.0%	100.0%	98.6%	73.8%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATP8A2	100.0%	100.0%	100.0%	99.5%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268
ATP9A	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242
ATR	100.0%	100.0%	100.0%	99.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	99.9%	99.7%	96.9%	68.4%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
ATXN2L	100.0%	100.0%	100.0%	99.4%	
AUH	100.0%	100.0%	100.0%	99.5%	3-methylglutaconic aciduria, type I, 250950
AUTS2	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVPR2	100.0%	100.0%	99.8%	86.7%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539

B3GALNT2	92.4%	92.4%	100.0%	99.1%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181
B3GALT6	99.9%	98.0%	100.0%	99.8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B3GLCT	100.0%	100.0%	100.0%	99.1%	Peters-plus syndrome, 261540
B4GALNT1	100.0%	100.0%	100.0%	99.7%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	100.0%	100.0%	100.0%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	100.0%	100.0%	100.0%	99.7%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100.0%	100.0%	100.0%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287
B9D1	100.0%	100.0%	100.0%	99.8%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100.0%	100.0%	100.0%	99.8%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BAP1	100.0%	100.0%	100.0%	99.8%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BAZ2B	100.0%	100.0%	100.0%	99.0%	
BBS1	100.0%	100.0%	100.0%	99.8%	Bardet-Biedl syndrome 1, 209900

BBS10	100.0%	100.0%	100.0%	99.7%	Bardet-Biedl syndrome 10, 615987
BBS12	100.0%	100.0%	100.0%	99.6%	Bardet-Biedl syndrome 12, 615989
BBS2	100.0%	100.0%	100.0%	99.4%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100.0%	100.0%	100.0%	99.5%	Bardet-Biedl syndrome 4, 615982
BBS5	100.0%	100.0%	100.0%	99.2%	Bardet-Biedl syndrome 5, 615983
BBS7	100.0%	100.0%	100.0%	99.3%	Bardet-Biedl syndrome 7, 615984
BBS9	95.8%	95.8%	100.0%	99.1%	Bardet-Biedl syndrome 9, 615986
BCAP31	99.1%	92.8%	99.2%	77.3%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100.0%	100.0%	100.0%	99.6%	Hengel-Maroofian-Schols syndrome, 619641
BCKDHA	100.0%	100.0%	100.0%	99.6%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100.0%	99.8%	100.0%	99.4%	Maple syrup urine disease, type Ib, 620698
BCKDK	100.0%	100.0%	100.0%	99.8%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCL11A	100.0%	100.0%	100.0%	99.6%	Dias-Logan syndrome, 617101
BCL11B	99.9%	99.6%	100.0%	99.7%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	100.0%	99.8%	98.6%	77.1%	Microphthalmia, syndromic 2, 300166
BCORL1	100.0%	99.5%	99.0%	74.3%	Shukla-Vernon syndrome, 301029

BCS1L	100.0%	100.0%	100.0%	99.9%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BICD2	100.0%	100.0%	100.0%	99.9%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BICRA	100.0%	100.0%	99.8%	97.0%	Coffin-Siris syndrome 12, 619325
BLM	100.0%	100.0%	100.0%	99.2%	Bloom syndrome, 210900
BLOC1S1	100.0%	100.0%	100.0%	98.7%	
BOLA3	100.0%	100.0%	100.0%	99.4%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BPTF	100.0%	100.0%	100.0%	98.6%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755
BRAF	100.0%	100.0%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRF1	100.0%	100.0%	100.0%	99.9%	Cerebellofaciodental syndrome, 616202

BRPF1	100.0%	99.9%	100.0%	99.6%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	100.0%	100.0%	100.0%	99.3%	
BRWD3	100.0%	99.7%	98.5%	74.1%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	100.0%	100.0%	100.0%	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	94.4%	94.3%	100.0%	99.5%	Biotinidase deficiency, 253260
BUB1	100.0%	100.0%	100.0%	99.5%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183
BUB1B	100.0%	100.0%	100.0%	99.5%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
C12orf4	100.0%	100.0%	100.0%	99.0%	Intellectual developmental disorder, autosomal recessive 66, 618221
C12orf57	100.0%	100.0%	100.0%	99.5%	Temtamy syndrome, 218340
C12orf65	100.0%	100.0%	100.0%	98.6%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
C2CD3	96.0%	96.0%	100.0%	99.4%	Orofaciodigital syndrome XIV, 615948

C2orf69	100.0%	100.0%	100.0%	99.3%	Combined oxidative phosphorylation deficiency 53, 619423
CA2	100.0%	100.0%	100.0%	99.5%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	100.0%	100.0%	100.0%	99.1%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100.0%	100.0%	100.0%	99.6%	Cerebellar ataxia, impaired intellectual development and dysequilibrium syndrome 3, 613227
CACNA1A	100.0%	100.0%	100.0%	99.2%	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	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497
CACNA1C	100.0%	100.0%	100.0%	99.4%	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	100.0%	100.0%	100.0%	99.3%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 69, 618285

CACNA1G	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1I	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114
CACNA2D1	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 110, 620149
CACNA2D2	100.0%	100.0%	100.0%	99.5%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 50, 616457
CAMK2A	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095
CAMK2B	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2G	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	99.9%	99.7%	100.0%	98.9%	
CAMSAP1	100.0%	100.0%	100.0%	99.6%	Cortical dysplasia, complex, with other brain malformations 12, 620316
CAMTA1	100.0%	100.0%	100.0%	99.3%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	100.0%	100.0%	100.0%	99.9%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719

CAPN15	100.0%	100.0%	100.0%	99.8%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPRIN1	100.0%	100.0%	100.0%	98.5%	Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636
CARS1	100.0%	100.0%	100.0%	99.8%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100.0%	100.0%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	100.0%	100.0%	98.7%	73.4%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CBL	100.0%	100.0%	100.0%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBS	100.0%	100.0%	100.0%	100.0%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CC2D1A	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 3, 608443
CC2D2A	98.2%	98.2%	100.0%	99.4%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285

CCBE1	100.0%	100.0%	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC115	100.0%	100.0%	100.0%	98.8%	Congenital disorder of glycosylation, type IIo, 616828
CCDC174	100.0%	100.0%	100.0%	98.6%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100.0%	100.0%	100.0%	98.4%	
CCDC22	100.0%	99.8%	98.9%	76.6%	Ritscher-Schinzel syndrome 2, 300963
CCDC32	100.0%	100.0%	100.0%	99.2%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC47	100.0%	100.0%	100.0%	98.9%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC88A	97.4%	97.4%	100.0%	97.7%	?PEHO syndrome-like, 617507
CCDC88C	100.0%	100.0%	100.0%	99.7%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600
CCND2	100.0%	100.0%	100.0%	99.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	99.4%	95.7%	97.1%	90.0%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147
CDC42	100.0%	100.0%	100.0%	99.4%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100.0%	100.0%	100.0%	99.7%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841
CDC6	100.0%	100.0%	100.0%	99.4%	?Meier-Gorlin syndrome 5, 613805
CDH11	100.0%	100.0%	100.0%	99.7%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380

CDH15	100.0%	100.0%	100.0%	99.9%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	100.0%	100.0%	100.0%	99.5%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDK10	100.0%	100.0%	100.0%	99.9%	Al Kaissi syndrome, 617694
CDK13	100.0%	100.0%	100.0%	99.5%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK19	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 87, 618916
CDK5RAP2	100.0%	100.0%	100.0%	99.4%	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	95.7%	95.3%	98.0%	71.6%	Developmental and epileptic encephalopathy 2, 300672
CDKN1C	100.0%	100.0%	100.0%	99.8%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDON	100.0%	100.0%	100.0%	99.5%	Holoprosencephaly 11, 614226
CELF2	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 97, 619561
CENPF	100.0%	100.0%	100.0%	98.8%	Stromme syndrome, 243605
CENPJ	100.0%	100.0%	100.0%	99.2%	Microcephaly 6, primary, autosomal recessive, 608393;?Seckel syndrome 4, 613676

CEP104	100.0%	100.0%	100.0%	99.0%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988
CEP120	100.0%	100.0%	100.0%	99.6%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP135	100.0%	100.0%	100.0%	98.8%	Microcephaly 8, primary, autosomal recessive, 614673
CEP152	100.0%	100.0%	100.0%	99.1%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP290	100.0%	100.0%	100.0%	98.5%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP41	100.0%	100.0%	100.0%	99.0%	Joubert syndrome 15, 614464
CEP55	100.0%	100.0%	100.0%	99.5%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100.0%	100.0%	99.9%	98.4%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	100.0%	100.0%	100.0%	98.9%	?Seckel syndrome 6, 614728
CEP83	100.0%	100.0%	100.0%	98.0%	Nephronophthisis 18, 615862
CEP85L	100.0%	100.0%	100.0%	99.1%	Lissencephaly 10, 618873
CEP89	100.0%	100.0%	100.0%	98.3%	
CERT1	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 34, 616351

CHAMP1	100.0%	100.0%	100.0%	98.2%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579
CHD1	100.0%	100.0%	100.0%	98.6%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100.0%	99.9%	100.0%	98.1%	Snijders Blok-Campeau syndrome, 618205
CHD4	100.0%	100.0%	100.0%	99.1%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100.0%	100.0%	100.0%	99.6%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHD7	100.0%	100.0%	100.0%	99.5%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHD8	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHKA	100.0%	100.0%	100.0%	97.9%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023
CHKB	100.0%	100.0%	100.0%	99.3%	Muscular dystrophy, congenital, megaconial type, 602541
CHMP1A	100.0%	100.0%	100.0%	99.9%	Pontocerebellar hypoplasia, type 8, 614961
CHRM1	100.0%	100.0%	100.0%	99.9%	
CHRNA4	100.0%	100.0%	99.9%	98.8%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513
CIC	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder, autosomal dominant 45, 617600

CIT	100.0%	100.0%	100.0%	99.4%	Microcephaly 17, primary, autosomal recessive, 617090
CKAP2L	100.0%	100.0%	100.0%	99.2%	Filippi syndrome, 272440
CLCN3	96.5%	96.5%	100.0%	99.4%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100.0%	100.0%	99.0%	74.7%	Raynaud-Claes syndrome, 300114
CLDN11	100.0%	100.0%	100.0%	99.9%	Leukodystrophy, hypomyelinating, 22, 619328
CLDN5	100.0%	100.0%	100.0%	99.9%	
CLIC2	100.0%	100.0%	98.2%	75.0%	?Intellectual developmental disorder, X-linked syndromic 32, 300886
CLIP1	100.0%	100.0%	100.0%	98.8%	
CLN3	93.2%	93.1%	100.0%	99.3%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83.1%	83.0%	100.0%	98.6%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100.0%	100.0%	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100.0%	100.0%	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100.0%	100.0%	100.0%	99.8%	Pontocerebellar hypoplasia, type 10, 615803

CLPB	100.0%	100.0%	100.0%	98.9%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLTC	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal dominant 56, 617854
CNKSR2	99.5%	98.5%	98.8%	73.4%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008
CNNM2	100.0%	100.0%	100.0%	99.6%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
CNOT1	100.0%	100.0%	100.0%	99.4%	Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNOT9	97.0%	91.9%	100.0%	99.6%	
CNPY3	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 60, 617929
CNTNAP1	100.0%	100.0%	100.0%	99.4%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100.0%	100.0%	100.0%	99.5%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100

COA8	100.0%	99.9%	100.0%	98.9%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100.0%	100.0%	100.0%	99.5%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COG1	100.0%	100.0%	100.0%	99.7%	Congenital disorder of glycosylation, type IIg, 611209
COG4	100.0%	100.0%	100.0%	99.5%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	100.0%	100.0%	100.0%	99.2%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100.0%	100.0%	100.0%	99.3%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	100.0%	100.0%	100.0%	99.5%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100.0%	100.0%	100.0%	99.7%	Congenital disorder of glycosylation, type IIh, 611182
COL18A1	100.0%	100.0%	100.0%	99.8%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880
COL4A1	100.0%	100.0%	100.0%	99.2%	?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	100.0%	100.0%	100.0%	99.6%	Brain small vessel disease 2, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519
COLEC11	100.0%	100.0%	100.0%	100.0%	3MC syndrome 2, 265050
COPB1	100.0%	100.0%	100.0%	98.9%	Baralle-Macken syndrome, 619255
COPB2	100.0%	100.0%	100.0%	99.5%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	96.3%	96.3%	100.0%	99.7%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100.0%	100.0%	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ8A	100.0%	100.0%	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100.0%	100.0%	100.0%	99.6%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100.0%	100.0%	99.9%	98.3%	Mitochondrial complex IV deficiency, nuclear type 3, 619046
COX15	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	100.0%	100.0%	100.0%	99.5%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX6B1	100.0%	100.0%	100.0%	99.9%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
CPE	100.0%	100.0%	100.0%	99.7%	BDV syndrome, 619326
CPLANE1	100.0%	100.0%	100.0%	99.3%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615

CPLX1	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 63, 617976
CPS1	100.0%	100.0%	100.0%	99.4%	Carbamoylphosphate synthetase I deficiency, 237300;{Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPSF3	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CRADD	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499
CRBN	100.0%	99.1%	100.0%	99.1%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREBBP	100.0%	100.0%	100.0%	99.2%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRLF1	99.7%	98.6%	99.7%	92.9%	Cold-induced sweating syndrome 1, 272430
CRPPA	100.0%	100.0%	100.0%	99.0%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSDE1	100.0%	100.0%	100.0%	99.5%	
CSF1R	100.0%	100.0%	100.0%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSNK1G1	100.0%	100.0%	100.0%	98.9%	
CSNK2A1	94.2%	94.2%	100.0%	99.6%	Okur-Chung neurodevelopmental syndrome, 617062

CSNK2B	100.0%	100.0%	100.0%	98.7%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	100.0%	100.0%	100.0%	99.2%	Joubert syndrome 21, 615636
CSTB	100.0%	100.0%	100.0%	99.7%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	100.0%	99.5%	100.0%	98.3%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100.0%	100.0%	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	100.0%	100.0%	100.0%	99.9%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	99.8%	99.4%	100.0%	99.6%	Cortical dysplasia, complex, with other brain malformations 9, 618174
CTNNB1	100.0%	100.0%	100.0%	99.6%	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
CTNND1	100.0%	100.0%	100.0%	99.3%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100.0%	99.9%	100.0%	98.9%	
CTR9	100.0%	100.0%	100.0%	99.2%	
CTSA	100.0%	100.0%	100.0%	99.3%	Galactosialidosis, 256540
CTSD	100.0%	100.0%	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 10, 610127

CTTNBP2	100.0%	100.0%	100.0%	99.4%	
CTU2	100.0%	100.0%	100.0%	99.9%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL3	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496
CUL4B	100.0%	99.9%	98.1%	72.0%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUX1	100.0%	100.0%	100.0%	99.0%	Global developmental delay with or without impaired intellectual development, 618330
CUX2	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 67, 618141
CWC27	100.0%	100.0%	100.0%	98.4%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100.0%	100.0%	100.0%	99.3%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CXorf56	100.0%	99.7%	98.0%	71.2%	?Intellectual developmental disorder, X-linked 107, 301013
CYB5R3	100.0%	100.0%	100.0%	99.7%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYFIP2	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 65, 618008
CYP27A1	100.0%	100.0%	100.0%	99.8%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100.0%	100.0%	100.0%	99.6%	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	100.0%	100.0%	100.0%	99.7%	D-2-hydroxyglutaric aciduria, 600721

DAG1	100.0%	100.0%	100.0%	99.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAGLA	100.0%	100.0%	100.0%	99.9%	
DARS1	100.0%	100.0%	100.0%	99.1%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100.0%	100.0%	100.0%	98.5%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	100.0%	100.0%	100.0%	99.2%	Maple syrup urine disease, type II, 620699
DCAF17	100.0%	100.0%	100.0%	99.8%	Woodhouse-Sakati syndrome, 241080
DCC	100.0%	100.0%	100.0%	99.5%	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
DCHS1	100.0%	100.0%	100.0%	99.9%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DCPS	100.0%	100.0%	100.0%	98.9%	Al-Raqad syndrome, 616459
DCX	98.9%	98.8%	98.5%	76.9%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DDB1	100.0%	100.0%	100.0%	99.7%	White-Kernohan syndrome, 619426
DDC	100.0%	100.0%	100.0%	99.3%	Aromatic L-amino acid decarboxylase deficiency, 608643

DDHD2	100.0%	100.0%	100.0%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	100.0%	100.0%	100.0%	99.9%	Warsaw breakage syndrome, 613398
DDX23	100.0%	100.0%	100.0%	99.1%	
DDX3X	99.1%	98.3%	98.9%	73.7%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DDX59	100.0%	100.0%	100.0%	99.0%	Orofaciodigital syndrome V, 174300
DDX6	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	100.0%	100.0%	100.0%	98.0%	Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100.0%	100.0%	100.0%	99.6%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100.0%	100.0%	100.0%	99.5%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504
DHCR24	100.0%	100.0%	100.0%	99.8%	Desmosterolosis, 602398
DHCR7	100.0%	100.0%	100.0%	99.9%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	94.4%	94.4%	100.0%	99.5%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861

DHFR	100.0%	100.0%	100.0%	99.6%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	96.7%	93.0%	100.0%	99.8%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHTKD1	100.0%	100.0%	100.0%	99.4%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX16	100.0%	100.0%	100.0%	99.3%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with variable motor and speech impairment, 617804
DHX37	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250
DHX9	100.0%	100.0%	100.0%	99.4%	
DIAPH1	100.0%	100.0%	100.0%	97.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIS3L2	100.0%	100.0%	100.0%	99.5%	Perlman syndrome, 267000
DKC1	100.0%	100.0%	98.0%	73.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLAT	100.0%	100.0%	100.0%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348

DLD	100.0%	100.0%	100.0%	99.3%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	100.0%	99.8%	99.1%	75.4%	Intellectual developmental disorder, X-linked 90, 300850
DLG4	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLL1	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DMD	99.5%	99.1%	98.5%	72.9%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DMPK	100.0%	100.0%	100.0%	99.6%	Myotonic dystrophy 1, 160900
DMXL2	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DNAJC12	100.0%	100.0%	100.0%	99.4%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100.0%	100.0%	100.0%	99.6%	3-methylglutaconic aciduria, type V, 610198
DNM1	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346
DNM1L	100.0%	100.0%	100.0%	99.5%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388

DNMT3A	100.0%	100.0%	100.0%	99.8%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	100.0%	100.0%	100.0%	99.6%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK3	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK6	100.0%	100.0%	100.0%	99.7%	Adams-Oliver syndrome 2, 614219
DOCK7	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 23, 615859
DOHH	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066
DOLK	100.0%	100.0%	100.0%	99.6%	Congenital disorder of glycosylation, type Im, 610768
DONSON	100.0%	100.0%	100.0%	99.8%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230
DPAGT1	100.0%	100.0%	100.0%	99.6%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPF2	100.0%	100.0%	100.0%	99.3%	Coffin-Siris syndrome 7, 618027
DPH1	100.0%	100.0%	100.0%	99.8%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901

DPH5	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070
DPM1	99.2%	96.6%	100.0%	98.6%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100.0%	100.0%	100.0%	99.3%	Congenital disorder of glycosylation, type Iu, 615042
DPP6	100.0%	99.9%	100.0%	99.1%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPYD	99.8%	99.6%	100.0%	99.4%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100.0%	100.0%	100.0%	99.5%	Dihydropyrimidinuria, 222748
DPYSL2	100.0%	100.0%	100.0%	99.2%	
DPYSL5	100.0%	100.0%	100.0%	99.5%	Ritscher-Schinzel syndrome 4, 619435
DTYMK	100.0%	100.0%	100.0%	99.9%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DYM	100.0%	99.9%	100.0%	99.0%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	100.0%	100.0%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492

DYRK1A	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal dominant 7, 614104
EARS2	100.0%	100.0%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100.0%	100.0%	100.0%	99.4%	Hypotonia, ataxia, and delayed development syndrome, 617330
EBP	100.0%	100.0%	99.2%	74.6%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	100.0%	100.0%	100.0%	99.0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	100.0%	100.0%	100.0%	99.8%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100.0%	100.0%	100.0%	99.3%	Congenital disorder of glycosylation, type IIv, 619493
EED	100.0%	100.0%	100.0%	98.4%	Cohen-Gibson syndrome, 617561
EEF1A2	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EEF1D	100.0%	100.0%	100.0%	99.5%	
EFNB2	100.0%	100.0%	100.0%	99.9%	
EFTUD2	100.0%	100.0%	100.0%	99.6%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EHMT1	100.0%	99.9%	100.0%	99.4%	Kleefstra syndrome 1, 610253
EIF2AK1	100.0%	100.0%	100.0%	99.0%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878

EIF2AK2	100.0%	100.0%	100.0%	98.5%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2AK3	100.0%	100.0%	100.0%	99.2%	Wolcott-Rallison syndrome, 226980
EIF2B4	100.0%	100.0%	100.0%	99.7%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF2S3	100.0%	100.0%	98.5%	73.7%	MEHMO syndrome, 300148
EIF3F	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 67, 618295
EIF4A2	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455
EIF4A3	100.0%	100.0%	100.0%	99.6%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	100.0%	100.0%	100.0%	99.4%	Faundes-Banka syndrome, 619376
ELAC2	100.0%	100.0%	100.0%	99.7%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELOVL4	100.0%	100.0%	100.0%	99.0%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELP2	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 58, 617270

EMC1	100.0%	100.0%	100.0%	99.2%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100.0%	100.0%	100.0%	99.2%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EML1	100.0%	100.0%	100.0%	99.3%	Band heterotopia, 600348
EMX2	100.0%	100.0%	100.0%	97.2%	Schizencephaly, 269160
ENTPD1	100.0%	100.0%	100.0%	99.5%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	100.0%	100.0%	100.0%	99.6%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
EPG5	100.0%	100.0%	100.0%	99.3%	Vici syndrome, 242840
EPHA7	100.0%	100.0%	100.0%	99.7%	
ERCC1	100.0%	100.0%	100.0%	99.5%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	100.0%	100.0%	100.0%	99.7%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100.0%	100.0%	100.0%	99.5%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651
ERCC5	100.0%	100.0%	100.0%	99.3%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

ERCC6	100.0%	100.0%	100.0%	99.4%	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
ERCC8	100.0%	100.0%	100.0%	99.4%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERI1	100.0%	100.0%	100.0%	98.8%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ERLIN2	100.0%	100.0%	100.0%	99.4%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ESAM	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371
ESCO2	100.0%	100.0%	100.0%	98.7%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
ETFB	100.0%	100.0%	100.0%	99.9%	Glutaric acidemia IIB, 231680
ETHE1	100.0%	100.0%	100.0%	99.3%	Ethylmalonic encephalopathy, 602473
EXOC2	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC7	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with seizures and brain atrophy, 619072

EXOSC2	100.0%	100.0%	100.0%	99.2%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100.0%	100.0%	100.0%	99.8%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100.0%	100.0%	100.0%	99.5%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100.0%	100.0%	99.9%	99.0%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100.0%	100.0%	100.0%	99.3%	Pontocerebellar hypoplasia, type 1D, 618065
EXTL3	100.0%	100.0%	100.0%	99.9%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
EZH2	100.0%	100.0%	100.0%	99.6%	Weaver syndrome, 277590
FA2H	100.0%	100.0%	100.0%	99.8%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	100.0%	100.0%	100.0%	99.7%	Leukodystrophy, hypomyelinating, 5, 610532
FAM149B1	100.0%	100.0%	100.0%	99.7%	Joubert syndrome 36, 618763
FAM20C	100.0%	100.0%	100.0%	99.7%	Raine syndrome, 259775
FAM50A	100.0%	100.0%	98.6%	73.6%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261
FAR1	100.0%	100.0%	100.0%	99.6%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100.0%	100.0%	100.0%	99.5%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSB	100.0%	100.0%	100.0%	99.3%	Rajab interstitial lung disease with brain calcifications 1, 613658

FAT4	99.9%	99.8%	100.0%	99.5%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBRSL1	99.9%	99.1%	100.0%	96.8%	
FBXL3	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	100.0%	100.0%	100.0%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100.0%	100.0%	100.0%	98.4%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO28	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 100, 619777
FBXO31	100.0%	100.0%	100.0%	99.5%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXW11	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW7	99.5%	98.2%	100.0%	99.4%	Developmental delay, hypotonia, and impaired language, 620012
FDFT1	100.0%	100.0%	100.0%	99.4%	Squalene synthase deficiency, 618156
FGD1	99.9%	99.5%	98.6%	75.4%	Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400
FGF12	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 47, 617166
FGF13	100.0%	99.8%	98.1%	74.3%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095

FGF14	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174
FGFR1	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	99.5%	Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400;Scaphocephaly and Axenfeld-Rieger anomaly, ;Craniosynostosis, nonspecific,

FGFR3	100.0%	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.2%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FIBP	100.0%	100.0%	100.0%	99.8%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	100.0%	100.0%	100.0%	99.5%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FIGN	100.0%	100.0%	100.0%	99.7%	
FILIP1	100.0%	100.0%	100.0%	98.8%	

FKRP	100.0%	100.0%	100.0%	99.9%	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
FKTN	100.0%	100.0%	100.0%	99.3%	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
FLNA	100.0%	99.9%	99.7%	83.8%	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
FLVCR1	100.0%	100.0%	100.0%	99.7%	Ataxia, posterior column, with retinitis pigmentosa, 609033

FLVCR2	100.0%	100.0%	100.0%	99.6%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	100.0%	99.8%	99.4%	93.7%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMR1	100.0%	100.0%	97.7%	71.5%	Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360
FOLR1	100.0%	100.0%	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOSL2	100.0%	100.0%	100.0%	99.5%	
FOXG1	100.0%	99.9%	100.0%	98.6%	Rett syndrome, congenital variant, 613454
FOXJ1	100.0%	100.0%	100.0%	99.8%	Ciliary dyskinesia, primary, 43, 618699
FOXP1	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder with language impairment with or without autistic features, 613670
FOXP2	100.0%	99.9%	100.0%	99.5%	Speech-language disorder-1, 602081
FOXRED1	100.0%	100.0%	100.0%	99.4%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRA10AC1	100.0%	100.0%	100.0%	98.8%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113
FRAS1	100.0%	99.9%	100.0%	99.6%	Fraser syndrome 1, 219000
FRMD4A	96.5%	96.5%	100.0%	99.3%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD5	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094

FRMPD4	100.0%	99.8%	98.2%	73.0%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100.0%	100.0%	100.0%	96.7%	Developmental and epileptic encephalopathy 37, 616981
FTCD	100.0%	100.0%	99.9%	99.1%	Glutamate formiminotransferase deficiency, 229100
FTO	94.5%	94.5%	100.0%	99.4%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	100.0%	100.0%	99.1%	79.1%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100.0%	100.0%	100.0%	99.4%	Fucosidosis, 230000
FUT8	100.0%	99.8%	100.0%	99.3%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FZR1	100.0%	100.0%	100.0%	100.0%	Developmental and epileptic encephalopathy 109, 620145
GABBR1	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502
GABBR2	99.9%	99.7%	100.0%	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	100.0%	100.0%	100.0%	99.5%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136

GABRA2	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780
GABRA3	100.0%	99.9%	98.7%	75.0%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRA5	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 92, 617829
GABRB3	100.0%	100.0%	100.0%	99.3%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113
GABRD	100.0%	100.0%	100.0%	98.1%	{Epilepsy, idiopathic generalized, 10}, 613060;{Epilepsy, juvenile myoclonic, susceptibility to}, 613060;{Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060
GABRG2	92.9%	92.6%	100.0%	99.6%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 89, 619124
GALC	100.0%	100.0%	100.0%	99.7%	Krabbe disease, 245200
GALE	100.0%	100.0%	100.0%	99.8%	Galactose epimerase deficiency, 230350
GALNT2	100.0%	100.0%	100.0%	99.3%	Congenital disorder of glycosylation, type IIt, 618885

GALT	100.0%	100.0%	100.0%	99.6%	Galactosemia, 230400
GAMT	100.0%	100.0%	100.0%	99.6%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2B	100.0%	100.0%	100.0%	99.2%	GAND syndrome, 615074
GATM	100.0%	100.0%	100.0%	99.6%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GCH1	100.0%	100.0%	100.0%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	100.0%	100.0%	100.0%	99.3%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDI1	100.0%	100.0%	99.2%	79.5%	Intellectual developmental disorder, X-linked 41, 300849
GEMIN5	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	100.0%	100.0%	100.0%	99.8%	Alexander disease, 203450
GFER	100.0%	100.0%	100.0%	99.8%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	100.0%	100.0%	100.0%	99.3%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100.0%	100.0%	100.0%	99.3%	Combined oxidative phosphorylation deficiency 39, 618397
GIGYF1	100.0%	100.0%	100.0%	99.9%	

GJA1	100.0%	100.0%	100.0%	99.3%	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
GJB1	100.0%	100.0%	99.7%	80.3%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	99.8%	98.7%	100.0%	98.7%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GK	100.0%	100.0%	98.2%	72.2%	Glycerol kinase deficiency, 307030
GLB1	100.0%	100.0%	100.0%	99.6%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	100.0%	100.0%	100.0%	99.6%	Glycine encephalopathy1, 605899
GLI2	100.0%	100.0%	100.0%	99.9%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GLI3	100.0%	100.0%	100.0%	99.8%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700

GLIS3	100.0%	100.0%	100.0%	99.4%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA2	99.5%	98.4%	98.6%	74.4%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076
GLS	100.0%	100.0%	100.0%	99.7%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412;?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100.0%	100.0%	100.0%	99.4%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100.0%	100.0%	100.0%	99.8%	Glutamine deficiency, congenital, 610015
GLYCTK	100.0%	100.0%	100.0%	99.7%	D-glyceric aciduria, 220120
GM2A	100.0%	100.0%	100.0%	99.8%	GM2-gangliosidosis, AB variant, 272750
GMNN	100.0%	100.0%	100.0%	99.7%	Meier-Gorlin syndrome 6, 616835
GMPPA	100.0%	100.0%	100.0%	99.6%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510
GMPPB	100.0%	100.0%	100.0%	99.8%	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

GNAI1	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854
GNAO1	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493
GNAS	100.0%	99.6%	100.0%	98.1%	ACTH-independent macronodular adrenal hyperplasia, 219080;Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;Pseudohypoparathyroidism Ib, 603233;McCune-Albright syndrome, somatic, mosaic, 174800;Pseudopseudohypoparathyroidism, 612463
GNB1	100.0%	100.0%	100.0%	99.7%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973
GNB2	100.0%	100.0%	100.0%	100.0%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNB5	100.0%	100.0%	100.0%	99.2%	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

GNPAT	100.0%	100.0%	100.0%	99.3%	Rhizomelic chondrodysplasia punctata, type 2, 222765
GNPTAB	100.0%	100.0%	100.0%	99.1%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	100.0%	100.0%	100.0%	99.5%	Mucolipidosis III gamma, 252605
GNS	100.0%	100.0%	100.0%	99.9%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100.0%	100.0%	100.0%	99.6%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GOT2	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 82, 618721
GPAA1	100.0%	100.0%	100.0%	99.9%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPC3	99.6%	98.9%	98.1%	72.5%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100.0%	99.8%	99.0%	75.9%	Keipert syndrome, 301026
GPHN	100.0%	99.9%	100.0%	99.3%	Molybdenum cofactor deficiency C, 615501
GPSM2	100.0%	100.0%	100.0%	99.5%	Chudley-McCullough syndrome, 604213
GPT2	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281
GRIA2	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	99.7%	99.2%	98.7%	72.8%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699
GRIA4	99.9%	99.8%	100.0%	99.5%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864

GRID2	99.9%	99.9%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	95.7%	95.5%	100.0%	99.4%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092
GRIN1	100.0%	100.0%	100.0%	99.5%	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
GRIN2A	99.8%	99.3%	100.0%	99.6%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	99.9%	99.8%	100.0%	99.7%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99.7%	98.7%	100.0%	97.8%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100.0%	100.0%	100.0%	99.5%	Fraser syndrome 3, 617667
GRM1	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRM7	100.0%	99.9%	100.0%	99.1%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922

GRN	100.0%	100.0%	100.0%	99.8%	Aphasia, primary progressive, 607485;Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
GSE1	100.0%	100.0%	100.0%	99.9%	
GSS	100.0%	100.0%	100.0%	99.7%	Hemolytic anemia due to glutathione synthetase deficiency, 231900;Glutathione synthetase deficiency, 266130
GTF2E2	100.0%	100.0%	100.0%	97.4%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	70.4%	70.3%	100.0%	99.5%	Trichothiodystrophy 3, photosensitive, 616395
GTPBP2	100.0%	100.0%	100.0%	99.3%	Jaberi-Elahi syndrome, 617988
GTPBP3	100.0%	100.0%	100.0%	99.7%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	100.0%	100.0%	100.0%	99.4%	Mucopolysaccharidosis VII, 253220
H1-4	100.0%	100.0%	100.0%	99.4%	Rahman syndrome, 617537
H3-3B	100.0%	100.0%	100.0%	99.7%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721
H4C3	100.0%	100.0%	100.0%	99.0%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758
H4C5	100.0%	100.0%	100.0%	98.3%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950
H4C9	100.0%	100.0%	100.0%	99.1%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951

HAAO	100.0%	100.0%	100.0%	99.8%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	100.0%	100.0%	100.0%	99.3%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100.0%	100.0%	100.0%	99.6%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	100.0%	100.0%	100.0%	99.3%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	100.0%	100.0%	100.0%	99.6%	Mitochondrial trifunctional protein deficiency 2, 620300
HAX1	100.0%	100.0%	100.0%	98.6%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	100.0%	100.0%	98.1%	72.8%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100.0%	99.9%	99.4%	80.5%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCN1	99.9%	99.7%	100.0%	99.2%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HDAC4	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	100.0%	99.9%	99.2%	78.0%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863

HDAC8	97.6%	97.2%	98.7%	73.3%	Cornelia de Lange syndrome 5, 300882
HEATR3	100.0%	100.0%	100.0%	99.2%	Diamond-Blackfan anemia 21, 620072
HEATR5B	100.0%	100.0%	100.0%	99.4%	
HECTD4	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250
HECW2	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HEPACAM	100.0%	100.0%	100.0%	99.5%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HERC1	100.0%	100.0%	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100.0%	99.9%	100.0%	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
HESX1	100.0%	100.0%	100.0%	97.4%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100.0%	100.0%	100.0%	99.9%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800

HEXB	100.0%	100.0%	100.0%	99.0%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	92.4%	92.4%	100.0%	99.6%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544
HIBCH	100.0%	100.0%	100.0%	98.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983
HIVEP2	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal dominant 43, 616977
HK1	100.0%	100.0%	100.0%	99.6%	Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547;Hemolytic anemia due to hexokinase deficiency, 235700
HLCS	100.0%	100.0%	100.0%	99.6%	Holocarboxylase synthetase deficiency, 253270
HMGB1	100.0%	100.0%	100.0%	98.7%	
HMGCL	100.0%	100.0%	100.0%	99.3%	HMG-CoA lyase deficiency, 246450
HNMT	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807
HNRNPD	100.0%	100.0%	100.0%	99.4%	
HNRNPH1	100.0%	100.0%	100.0%	98.8%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083

HNRNPH2	100.0%	100.0%	99.2%	76.0%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986
HNRNPK	100.0%	100.0%	100.0%	99.8%	Au-Kline syndrome, 616580
HNRNPU	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 54, 617391
HOXA1	100.0%	100.0%	100.0%	99.4%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536
HPD	100.0%	100.0%	100.0%	99.1%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	100.0%	100.0%	98.5%	75.3%	Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322
HRAS	100.0%	100.0%	100.0%	99.7%	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
HS2ST1	100.0%	100.0%	100.0%	99.5%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HSD17B10	100.0%	99.8%	99.6%	75.5%	HSD10 mitochondrial disease, 300438

HSD17B4	96.6%	96.6%	100.0%	99.3%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSPA9	100.0%	100.0%	100.0%	99.4%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPD1	100.0%	100.0%	100.0%	99.2%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100.0%	100.0%	100.0%	99.3%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248
HUWE1	100.0%	99.8%	98.7%	76.2%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
HYLS1	100.0%	100.0%	100.0%	99.9%	Hydrolethalus syndrome, 236680
IARS1	100.0%	100.0%	100.0%	99.5%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100.0%	100.0%	100.0%	99.5%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100.0%	100.0%	100.0%	100.0%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IDS	100.0%	100.0%	98.5%	72.9%	Mucopolysaccharidosis II, 309900
IDUA	100.0%	100.0%	100.0%	99.9%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IER3IP1	100.0%	100.0%	100.0%	99.6%	Microcephaly, epilepsy, and diabetes syndrome, 614231

IFIH1	100.0%	100.0%	100.0%	99.0%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IFT140	100.0%	100.0%	100.0%	99.6%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781
IFT172	100.0%	100.0%	100.0%	99.4%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100.0%	100.0%	100.0%	99.4%	Bardet-Biedl syndrome 19, 615996
IFT74	100.0%	100.0%	100.0%	99.1%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT81	94.9%	94.9%	100.0%	98.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	100.0%	99.9%	97.5%	70.6%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100.0%	100.0%	100.0%	99.0%	Insulin-like growth factor I deficiency, 608747
IGF1R	100.0%	100.0%	100.0%	99.7%	Insulin-like growth factor I, resistance to, 270450
IKBKG	99.9%	98.4%	99.1%	80.1%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IL1RAPL1	100.0%	100.0%	97.9%	73.7%	Intellectual developmental disorder, X-linked 21, 300143

IMPA1	100.0%	100.0%	100.0%	98.7%	Intellectual developmental disorder, autosomal recessive 59, 617323
INPP5E	100.0%	100.0%	100.0%	99.8%	Joubert syndrome 1, 213300;Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INPP5K	100.0%	100.0%	100.0%	99.6%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404
INTS1	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS11	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
IPO8	100.0%	100.0%	100.0%	99.3%	VISS syndrome, 619472
IQSEC1	100.0%	99.9%	100.0%	98.6%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99.7%	98.4%	97.1%	69.8%	Intellectual developmental disorder, X-linked 1, 309530
IREB2	100.0%	100.0%	100.0%	99.5%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF2BPL	100.0%	100.0%	100.0%	97.0%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRX5	100.0%	100.0%	100.0%	98.8%	Hamamy syndrome, 611174
ISCA2	100.0%	100.0%	100.0%	99.6%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITGA7	100.0%	100.0%	100.0%	99.7%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204

ITPA	100.0%	100.0%	100.0%	99.0%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
ITPR1	100.0%	100.0%	100.0%	99.2%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
IVD	100.0%	100.0%	100.0%	99.8%	Isovaleric acidemia, 243500
JAG1	100.0%	100.0%	100.0%	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	100.0%	99.9%	100.0%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAM3	100.0%	100.0%	100.0%	99.8%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JARID2	100.0%	100.0%	100.0%	99.6%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098
JMJD1C	100.0%	100.0%	100.0%	98.9%	
KANK1	100.0%	100.0%	100.0%	99.5%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	100.0%	100.0%	100.0%	99.7%	Koolen-De Vries syndrome, 610443
KAT5	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100.0%	100.0%	100.0%	99.1%	Arboleda-Tham syndrome, 616268

KAT6B	100.0%	100.0%	100.0%	99.3%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KAT8	100.0%	100.0%	99.9%	98.6%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100.0%	100.0%	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA2	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100.0%	100.0%	100.0%	99.6%	
KCNA4	100.0%	100.0%	100.0%	99.1%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNB1	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100.0%	100.0%	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC2	100.0%	100.0%	100.0%	99.1%	Developmental and epileptic encephalopathy 103, 619913
KCNC3	99.7%	98.3%	99.7%	93.1%	Spinocerebellar ataxia 13, 605259
KCNH1	98.5%	98.5%	100.0%	99.5%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNH5	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 112, 620537
KCNJ10	100.0%	100.0%	100.0%	99.6%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780

KCNJ11	100.0%	100.0%	100.0%	99.9%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ6	100.0%	100.0%	100.0%	99.9%	Keppen-Lubinsky syndrome, 614098
KCNK3	100.0%	100.0%	100.0%	99.7%	Pulmonary hypertension, primary, 4, 615344
KCNK4	100.0%	100.0%	100.0%	99.6%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	100.0%	100.0%	100.0%	99.3%	Birk-Barel syndrome, 612292
KCNMA1	100.0%	99.9%	100.0%	99.2%	{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
KCNN2	99.9%	99.7%	100.0%	99.3%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCNN3	100.0%	100.0%	100.0%	99.4%	Zimmermann-Laband syndrome 3, 618658
KCNQ2	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200

KCNQ3	100.0%	100.0%	100.0%	99.3%	Seizures, benign neonatal, 2, 121201
KCNQ5	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder, autosomal dominant 46, 617601
KCNT1	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	99.7%	99.4%	100.0%	99.5%	Developmental and epileptic encephalopathy 57, 617771
KCTD3	100.0%	100.0%	100.0%	98.4%	
KCTD7	100.0%	100.0%	100.0%	99.9%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	100.0%	100.0%	100.0%	99.3%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM2B	100.0%	100.0%	100.0%	99.4%	
KDM3B	100.0%	100.0%	100.0%	99.5%	Diets-Jongmans syndrome, 618846
KDM4B	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5A	100.0%	100.0%	100.0%	99.4%	
KDM5B	97.5%	96.3%	100.0%	99.4%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	100.0%	99.9%	98.6%	75.4%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6A	100.0%	99.9%	98.5%	73.8%	Kabuki syndrome 2, 300867
KDM6B	100.0%	100.0%	100.0%	98.6%	Stolerman neurodevelopmental syndrome, 618505
KIAA0586	95.6%	95.5%	100.0%	99.0%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490

KIAA1109	100.0%	99.9%	100.0%	99.4%	Alkuraya-Kucinskas syndrome, 617822
KIDINS220	100.0%	100.0%	100.0%	99.4%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF11	100.0%	100.0%	100.0%	99.3%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KIF14	100.0%	100.0%	100.0%	98.9%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF1A	100.0%	100.0%	100.0%	99.9%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 610357
KIF21B	100.0%	100.0%	100.0%	99.6%	
KIF26A	100.0%	100.0%	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 11, 620156
KIF2A	100.0%	100.0%	100.0%	98.7%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100.0%	100.0%	100.0%	98.5%	Retinitis pigmentosa 89, 618955
KIF4A	100.0%	100.0%	98.7%	73.5%	Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923
KIF5A	100.0%	100.0%	100.0%	98.9%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187

KIF5C	99.3%	99.3%	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100.0%	99.9%	100.0%	99.4%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	95.6%	95.6%	100.0%	99.4%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL3	100.0%	100.0%	100.0%	98.0%	
KLF7	100.0%	100.0%	100.0%	99.3%	
KLHL15	100.0%	100.0%	98.5%	72.2%	Intellectual developmental disorder, X-linked 103, 300982
KLHL20	100.0%	100.0%	100.0%	99.6%	
KMT2A	100.0%	100.0%	100.0%	99.1%	Wiedemann-Steiner syndrome, 605130
KMT2B	99.8%	99.5%	99.9%	98.3%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KMT2C	100.0%	100.0%	100.0%	99.3%	Kleefstra syndrome 2, 617768
KMT2D	100.0%	100.0%	100.0%	99.6%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KMT2E	100.0%	99.9%	100.0%	98.7%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNL1	98.7%	98.7%	100.0%	99.4%	Microcephaly 4, primary, autosomal recessive, 604321

KPTN	100.0%	100.0%	99.9%	99.2%	Intellectual developmental disorder, autosomal recessive 41, 615637
KRAS	100.0%	100.0%	100.0%	99.8%	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
L1CAM	100.0%	99.9%	99.4%	79.3%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
L2HGDH	100.0%	100.0%	100.0%	99.4%	L-2-hydroxyglutaric aciduria, 236792
LAMA1	100.0%	100.0%	100.0%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMA2	99.8%	99.5%	100.0%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	100.0%	100.0%	100.0%	99.4%	Lissencephaly 5, 615191
LAMB2	100.0%	100.0%	100.0%	99.9%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049

LAMC3	100.0%	100.0%	100.0%	99.6%	Cortical malformations, occipital, 614115
LAMP2	100.0%	100.0%	98.7%	75.7%	Danon disease, 300257
LARGE1	100.0%	100.0%	100.0%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154
LARP7	100.0%	100.0%	100.0%	98.5%	Alazami syndrome, 615071
LARS1	100.0%	100.0%	100.0%	99.3%	?Infantile liver failure syndrome 1, 615438
LAS1L	100.0%	99.9%	99.2%	76.0%	Wilson-Turner syndrome, 309585
LETM1	100.0%	100.0%	100.0%	99.7%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LHX2	100.0%	100.0%	100.0%	99.3%	
LIAS	100.0%	100.0%	100.0%	99.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	100.0%	100.0%	100.0%	99.4%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LINGO1	100.0%	100.0%	100.0%	100.0%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	100.0%	100.0%	100.0%	98.7%	Intellectual developmental disorder, autosomal recessive 27, 614340
LMAN2L	100.0%	100.0%	100.0%	99.2%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887

LMBRD2	100.0%	100.0%	100.0%	99.2%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMNB1	100.0%	100.0%	100.0%	99.4%	Leukodystrophy, adult-onset, autosomal dominant, 169500;Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	100.0%	99.8%	100.0%	99.4%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709
LONP1	100.0%	100.0%	100.0%	99.8%	CODAS syndrome, 600373
LRP2	100.0%	100.0%	100.0%	99.6%	Donnai-Barrow syndrome, 222448
LRPPRC	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LSS	100.0%	100.0%	100.0%	99.9%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840
LYRM7	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	100.0%	99.8%	100.0%	99.4%	Chediak-Higashi syndrome, 214500
LZTFL1	100.0%	100.0%	100.0%	98.9%	Bardet-Biedl syndrome 17, 615994
LZTR1	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAB21L1	100.0%	100.0%	100.0%	97.7%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100.0%	100.0%	100.0%	100.0%	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877

MACF1	100.0%	100.0%	100.0%	99.3%	Lissencephaly 9 with complex brainstem malformation, 618325
MADD	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004
MAF	93.9%	89.9%	99.8%	95.0%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088
MAG	100.0%	100.0%	100.0%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	100.0%	100.0%	100.0%	99.8%	Schaaf-Yang syndrome, 615547
MAN1B1	100.0%	100.0%	100.0%	99.9%	Rafiq syndrome, 614202
MAN2B1	100.0%	100.0%	100.0%	99.7%	Mannosidosis, alpha-, types I and II, 248500
MAN2C1	100.0%	100.0%	100.0%	99.8%	Congenital disorder of deglycosylation 2, 619775
MANBA	100.0%	100.0%	100.0%	99.5%	Mannosidosis, beta, 248510
MAOA	99.4%	98.5%	98.4%	76.2%	{Antisocial behavior}, 300615;Brunner syndrome, 300615
MAP1B	100.0%	100.0%	100.0%	98.1%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP2K1	100.0%	100.0%	100.0%	99.3%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100.0%	100.0%	100.0%	99.8%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100.0%	100.0%	100.0%	99.3%	Noonan syndrome 13, 619087
MAPK8IP3	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443

MAPKAPK5	100.0%	100.0%	100.0%	99.2%	Neurocardiofaciodigital syndrome, 619869
MAPRE2	100.0%	100.0%	100.0%	99.6%	Symmetric circumferential skin creases, congenital, 2, 616734
MARS1	100.0%	100.0%	100.0%	99.6%	Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MASP1	100.0%	100.0%	100.0%	99.8%	3MC syndrome 1, 257920
MAST1	100.0%	100.0%	100.0%	99.8%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAST3	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 108, 620115
MAST4	100.0%	100.0%	100.0%	99.3%	
MAT1A	100.0%	100.0%	100.0%	99.9%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MBD5	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBOAT7	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 57, 617188

MBTPS2	100.0%	100.0%	99.4%	75.3%	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
MCCC1	100.0%	100.0%	100.0%	99.6%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	100.0%	100.0%	100.0%	98.6%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCM3AP	100.0%	100.0%	100.0%	99.6%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCOLN1	100.0%	100.0%	100.0%	99.9%	Mucolipidosis IV, 252650
MCPH1	100.0%	100.0%	100.0%	99.2%	Microcephaly 1, primary, autosomal recessive, 251200
MDH2	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 51, 617339
MECP2	100.0%	99.7%	99.1%	75.4%	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	100.0%	100.0%	100.0%	99.6%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629

MED11	100.0%	100.0%	100.0%	99.1%	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327
MED12	100.0%	99.8%	98.2%	71.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED12L	100.0%	100.0%	100.0%	99.4%	Nizon-Isidor syndrome, 618872
MED13	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	100.0%	99.6%	100.0%	99.5%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED17	100.0%	100.0%	100.0%	99.1%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100.0%	100.0%	100.0%	99.7%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MED27	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	100.0%	100.0%	100.0%	99.6%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEGF8	100.0%	100.0%	99.9%	99.0%	Carpenter syndrome 2, 614976

MEIS2	100.0%	100.0%	100.0%	99.7%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
METTL23	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100.0%	100.0%	100.0%	98.8%	Intellectual developmental disorder, autosomal recessive 72, 618665
MFF	100.0%	100.0%	100.0%	99.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100.0%	100.0%	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100.0%	100.0%	100.0%	99.5%	Congenital disorder of glycosylation, type IIa, 212066
MGP	100.0%	100.0%	100.0%	99.1%	Keutel syndrome, 245150
MIA3	100.0%	100.0%	100.0%	98.6%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MICU1	100.0%	99.9%	100.0%	99.7%	Myopathy with extrapyramidal signs, 615673
MID1	99.6%	99.1%	98.6%	76.3%	Opitz GBBB syndrome, 300000
MID2	100.0%	99.9%	99.1%	75.6%	?Intellectual developmental disorder, X-linked 101, 300928
MINPP1	100.0%	100.0%	100.0%	98.6%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527

MKKS	100.0%	100.0%	100.0%	99.6%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	100.0%	100.0%	100.0%	99.8%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MLC1	100.0%	100.0%	100.0%	99.8%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLYCD	100.0%	100.0%	100.0%	99.9%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100.0%	100.0%	100.0%	99.1%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	100.0%	100.0%	100.0%	99.0%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100.0%	100.0%	100.0%	99.2%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	89.3%	89.3%	100.0%	99.0%	Methylmalonic aciduria, cblD type, variant 2, 277410;Methylmalonic aciduria and homocystinuria, cblD type, 277410;Homocystinuria, cblD type, variant 1, 277410
MMGT1	100.0%	100.0%	98.7%	75.4%	
MMUT	100.0%	100.0%	100.0%	99.0%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100.0%	100.0%	100.0%	100.0%	CEBALID syndrome, 618774;Meningioma, 607174
MOCS1	100.0%	100.0%	100.0%	99.4%	Molybdenum cofactor deficiency A, 252150
MOCS2	100.0%	100.0%	100.0%	99.7%	Molybdenum cofactor deficiency B, 252160
MOGS	100.0%	100.0%	100.0%	99.9%	Congenital disorder of glycosylation, type IIb, 606056

MORC2	100.0%	100.0%	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPDU1	100.0%	100.0%	100.0%	97.7%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99.5%	99.1%	100.0%	99.3%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	100.0%	100.0%	100.0%	99.1%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPP5	100.0%	100.0%	100.0%	99.6%	
MPV17	100.0%	100.0%	100.0%	99.7%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRAS	100.0%	100.0%	100.0%	99.7%	Noonan syndrome 11, 618499
MRPS22	100.0%	100.0%	100.0%	98.9%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS34	100.0%	100.0%	100.0%	99.9%	Combined oxidative phosphorylation deficiency 32, 617664
MRTFB	100.0%	100.0%	100.0%	99.6%	
MSL2	100.0%	100.0%	100.0%	99.6%	
MSL3	100.0%	100.0%	98.6%	74.4%	Basilicata-Akhtar syndrome, 301032
MSMO1	100.0%	100.0%	100.0%	99.3%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MTFMT	100.0%	100.0%	100.0%	99.4%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248

MTHFR	100.0%	100.0%	100.0%	99.7%	Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634;{Vascular disease, susceptibility to},
MTHFS	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTO1	93.7%	91.1%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100.0%	100.0%	100.0%	99.7%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MTR	100.0%	100.0%	100.0%	99.3%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria-megaloblastic anemia, cblG complementation type, 250940
MTRR	100.0%	100.0%	100.0%	98.9%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTSS2	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086
MVK	90.4%	90.4%	100.0%	100.0%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYCN	100.0%	100.0%	100.0%	98.6%	Feingold syndrome 1, 164280

MYH9	100.0%	100.0%	100.0%	99.6%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO5A	100.0%	100.0%	100.0%	99.3%	Griscelli syndrome, type 1, 214450
MYO9A	100.0%	100.0%	100.0%	99.4%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYT1L	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal dominant 39, 616521
NAA10	100.0%	100.0%	98.9%	74.0%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NAA15	96.6%	96.6%	100.0%	99.0%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 73, 619717
NACC1	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAE1	100.0%	100.0%	100.0%	99.0%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210
NAGA	100.0%	100.0%	100.0%	99.7%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241
NAGLU	100.0%	100.0%	100.0%	99.9%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920

NALCN	100.0%	100.0%	100.0%	99.4%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	100.0%	100.0%	99.9%	99.1%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NAPB	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 107, 620033
NARS1	100.0%	100.0%	100.0%	99.4%	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	100.0%	100.0%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAXE	100.0%	100.0%	100.0%	99.7%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	99.7%	99.2%	100.0%	99.6%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBN	100.0%	100.0%	100.0%	98.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCAPG2	100.0%	100.0%	100.0%	99.4%	Khan-Khan-Katsanis syndrome, 618460

NCDN	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCKAP1	100.0%	100.0%	100.0%	99.3%	
NDE1	100.0%	100.0%	100.0%	99.2%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019
NDP	100.0%	100.0%	99.3%	80.1%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NDST1	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100.0%	100.0%	97.0%	73.1%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100.0%	98.8%	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	100.0%	100.0%	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA8	100.0%	100.0%	100.0%	99.8%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFAF3	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	100.0%	100.0%	100.0%	98.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100.0%	100.0%	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100.0%	100.0%	100.0%	99.1%	Mitochondrial complex I deficiency, nuclear type 5, 618226

NDUFS2	100.0%	100.0%	100.0%	99.2%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	96.6%	91.3%	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100.0%	99.9%	100.0%	99.1%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100.0%	100.0%	99.9%	99.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFS7	100.0%	100.0%	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100.0%	100.0%	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100.0%	100.0%	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	100.0%	100.0%	99.9%	98.6%	Periventricular nodular heterotopia 7, 617201
NEMF	100.0%	100.0%	100.0%	98.8%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEU1	100.0%	100.0%	100.0%	99.3%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEUROD2	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 72, 618374
NEUROG1	100.0%	100.0%	100.0%	99.9%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469

NEXMIF	100.0%	99.9%	98.1%	71.7%	Intellectual developmental disorder, X-linked 98, 300912
NF1	100.0%	100.0%	100.0%	99.3%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFE2L2	100.0%	100.0%	100.0%	99.6%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	100.0%	100.0%	100.0%	96.3%	Brain malformations with or without urinary tract defects, 613735
NFIB	100.0%	100.0%	100.0%	99.7%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100.0%	99.7%	99.9%	98.6%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NFU1	100.0%	100.0%	100.0%	99.4%	Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	100.0%	100.0%	100.0%	99.3%	Congenital disorder of deglycosylation 1, 615273
NHLRC2	100.0%	99.9%	100.0%	99.6%	FINCA syndrome, 618278
NHS	100.0%	100.0%	98.7%	74.1%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350
NIPBL	100.0%	100.0%	100.0%	99.4%	Cornelia de Lange syndrome 1, 122470
NKAP	100.0%	100.0%	98.5%	73.7%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039

NKX2-1	100.0%	100.0%	100.0%	99.8%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NLGN2	100.0%	100.0%	100.0%	99.3%	
NLGN3	100.0%	100.0%	98.9%	76.1%	{Autism susceptibility, X-linked 1}, 300425
NLGN4X	100.0%	99.9%	98.7%	76.5%	Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495
NONO	100.0%	99.5%	98.3%	75.7%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOVA2	100.0%	100.0%	100.0%	98.2%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPC1	100.0%	100.0%	100.0%	99.5%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	100.0%	100.0%	100.0%	98.9%	Niemann-pick disease, type C2, 607625
NPHP1	100.0%	100.0%	100.0%	99.4%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NR2F1	100.0%	99.9%	100.0%	98.5%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100.0%	100.0%	100.0%	99.3%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR4A2	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911

NRAS	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833
NRROS	100.0%	100.0%	100.0%	99.8%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	99.8%	99.7%	100.0%	99.8%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332
NSD1	100.0%	100.0%	100.0%	99.3%	Sotos syndrome, 117550
NSD2	100.0%	100.0%	100.0%	99.4%	Rauch-Steindl syndrome, 619695
NSDHL	100.0%	99.9%	98.9%	79.0%	CK syndrome, 300831;CHILD syndrome, 308050
NSF	100.0%	100.0%	99.9%	92.3%	Developmental and epileptic encephalopathy 96, 619340
NSRP1	91.0%	91.0%	100.0%	98.6%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001
NSUN2	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal recessive 5, 611091

NT5C2	100.0%	100.0%	100.0%	99.4%	Spastic paraplegia 45, autosomal recessive, 613162
NTNG2	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100.0%	100.0%	100.0%	99.6%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886
NUBPL	100.0%	100.0%	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDT2	100.0%	100.0%	100.0%	98.5%	Intellectual developmental disorder with or without peripheral neuropathy, 619844
NUP107	100.0%	100.0%	100.0%	99.0%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	100.0%	100.0%	100.0%	99.3%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP188	100.0%	100.0%	100.0%	99.6%	Sandestig-Stefanova syndrome, 618804
NUP214	100.0%	100.0%	100.0%	99.4%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426
NUP54	100.0%	100.0%	100.0%	99.5%	Dystonia 37, early-onset, with striatal lesions, 620427

NUP62	100.0%	100.0%	100.0%	99.9%	Striatonigral degeneration, infantile, 271930
NUP85	100.0%	100.0%	100.0%	98.9%	Nephrotic syndrome, type 17, 618176
NUS1	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
OAT	100.0%	100.0%	100.0%	99.6%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	100.0%	100.0%	99.9%	97.1%	Pseudo-TORCH syndrome 1, 251290
OCRL	100.0%	100.0%	98.2%	72.6%	Dent disease 2, 300555;Lowe syndrome, 309000
ODC1	100.0%	100.0%	100.0%	99.2%	Bachmann-Bupp syndrome, 619075
OFD1	100.0%	100.0%	97.9%	69.5%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDH	100.0%	100.0%	100.0%	99.7%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100.0%	100.0%	100.0%	99.7%	Yoon-Bellen neurodevelopmental syndrome, 619701
OGT	100.0%	99.9%	98.8%	77.1%	Intellectual developmental disorder, X-linked 106, 300997
OPA3	100.0%	100.0%	100.0%	99.8%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPHN1	100.0%	99.9%	98.5%	72.8%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486

ORC1	100.0%	100.0%	100.0%	99.6%	Meier-Gorlin syndrome 1, 224690
OSGEP	100.0%	100.0%	100.0%	99.7%	Galloway-Mowat syndrome 3, 617729
OTC	100.0%	99.6%	97.9%	72.9%	Ornithine transcarbamylase deficiency, 311250
OTUD5	100.0%	99.6%	98.5%	72.9%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100.0%	100.0%	100.0%	99.0%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	99.6%	98.3%	99.9%	97.2%	
OTX2	100.0%	100.0%	100.0%	99.6%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OXR1	100.0%	100.0%	100.0%	99.5%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000
P4HTM	100.0%	100.0%	100.0%	99.4%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPC1	100.0%	100.0%	100.0%	99.4%	
PACS1	100.0%	100.0%	100.0%	99.5%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100.0%	100.0%	100.0%	99.3%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432

PAH	100.0%	100.0%	100.0%	99.7%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PAK1	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	99.8%	99.3%	98.3%	70.4%	Intellectual developmental disorder, X-linked 30, 300558
PAM16	85.2%	84.5%	100.0%	99.8%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320
PAN2	100.0%	100.0%	100.0%	99.6%	
PANK2	100.0%	100.0%	100.0%	99.6%	HARP syndrome, 607236;Neurodegeneration with brain iron accumulation 1, 234200
PANX1	100.0%	100.0%	100.0%	99.6%	Oocyte/zygote/embryo maturation arrest 7, 618550
PARN	97.0%	95.9%	100.0%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP6	100.0%	100.0%	100.0%	99.7%	
PAX1	100.0%	100.0%	100.0%	99.2%	Otofaciocervical syndrome 2, 615560
PAX6	100.0%	100.0%	100.0%	98.4%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;?Coloboma, ocular, 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	100.0%	100.0%	100.0%	99.5%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220

PAX8	100.0%	100.0%	100.0%	99.4%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	100.0%	99.9%	100.0%	99.6%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	100.0%	100.0%	100.0%	99.8%	Pyruvate carboxylase deficiency, 266150
PCCA	100.0%	100.0%	100.0%	99.1%	Propionicacidemia, 606054
PCCB	99.9%	98.0%	100.0%	99.5%	Propionicacidemia, 606054
PCDH12	100.0%	100.0%	100.0%	99.8%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH19	100.0%	99.9%	98.8%	75.9%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880
PCGF2	100.0%	100.0%	100.0%	99.2%	Turnpenny-Fry syndrome, 618371
PCLO	99.9%	99.7%	100.0%	98.3%	?Pontocerebellar hypoplasia, type 3, 608027
PCNT	100.0%	100.0%	100.0%	99.6%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT2	100.0%	100.0%	100.0%	99.8%	Spastic paraplegia 82, autosomal recessive, 618770
PDE2A	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE4D	100.0%	99.9%	100.0%	99.1%	Acrodysostosis 2, with or without hormone resistance, 614613

PDGFRB	100.0%	100.0%	100.0%	99.8%	Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440
PDHA1	99.7%	97.5%	99.1%	75.1%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100.0%	100.0%	100.0%	99.2%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100.0%	99.8%	100.0%	99.4%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100.0%	100.0%	100.0%	99.9%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100.0%	100.0%	100.0%	99.6%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100.0%	100.0%	100.0%	99.3%	Coenzyme Q10 deficiency, primary, 3, 614652
PDZD8	100.0%	100.0%	100.0%	98.8%	Intellectual developmental disorder with autism and dysmorphic facies, 620021
PEPD	100.0%	100.0%	100.0%	99.9%	Prolidase deficiency, 170100
PET100	100.0%	100.0%	100.0%	99.8%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100.0%	100.0%	100.0%	99.3%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100.0%	100.0%	100.0%	100.0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871

PEX11B	100.0%	100.0%	100.0%	97.5%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100.0%	100.0%	100.0%	99.4%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100.0%	100.0%	100.0%	99.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX16	100.0%	100.0%	100.0%	99.3%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100.0%	100.0%	100.0%	99.6%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100.0%	100.0%	100.0%	99.6%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	100.0%	100.0%	100.0%	99.2%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100.0%	100.0%	100.0%	98.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100.0%	100.0%	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716

PEX6	100.0%	100.0%	100.0%	99.5%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	91.2%	91.2%	100.0%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGAP1	100.0%	100.0%	100.0%	99.2%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100.0%	100.0%	100.0%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207
PGAP3	100.0%	100.0%	100.0%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGK1	100.0%	99.7%	98.9%	73.6%	Phosphoglycerate kinase 1 deficiency, 300653
PGM2L1	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100.0%	100.0%	100.0%	99.6%	Immunodeficiency 23, 615816
PHACTR1	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 70, 618298
PHF21A	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	100.0%	100.0%	98.9%	75.6%	Borjeson-Forssman-Lehmann syndrome, 301900

PHF8	100.0%	99.9%	98.6%	73.4%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263
PHGDH	100.0%	100.0%	100.0%	99.8%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	100.0%	99.8%	100.0%	98.8%	Chung-Jansen syndrome, 617991
PI4KA	100.0%	99.8%	100.0%	99.5%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	100.0%	100.0%	100.0%	97.1%	Joubert syndrome 33, 617767
PIDD1	100.0%	100.0%	100.0%	99.9%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827
PIGA	100.0%	100.0%	98.6%	74.5%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100.0%	100.0%	100.0%	98.9%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100.0%	100.0%	100.0%	100.0%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100.0%	100.0%	100.0%	99.7%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356

PIGG	100.0%	100.0%	100.0%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	80.9%	75.0%	100.0%	99.5%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100.0%	100.0%	100.0%	98.8%	CHIME syndrome, 280000
PIGN	100.0%	99.9%	100.0%	99.3%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100.0%	100.0%	100.0%	99.9%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100.0%	100.0%	100.0%	99.9%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100.0%	100.0%	100.0%	99.3%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100.0%	100.0%	100.0%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300

PIGW	100.0%	100.0%	100.0%	99.6%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100.0%	100.0%	100.0%	99.3%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809
PIK3CA	100.0%	100.0%	100.0%	99.4%	Hemifacial myohyperplasia, somatic, 606733;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;Nevus, epidermal, somatic, 162900;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3R2	100.0%	100.0%	100.0%	99.3%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIP5K1C	100.0%	100.0%	100.0%	99.4%	Lethal congenital contractural syndrome 3, 611369
PISD	100.0%	100.0%	100.0%	99.9%	Liberfarb syndrome, 618889
PITRM1	100.0%	100.0%	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PJA1	100.0%	99.9%	97.6%	68.0%	

PLA2G6	100.0%	99.9%	100.0%	99.5%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLAA	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLCB1	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 12, 613722
PLK1	100.0%	100.0%	100.0%	99.4%	
PLK4	100.0%	100.0%	100.0%	99.1%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	99.9%	98.9%	98.5%	73.5%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PLPBP	100.0%	100.0%	100.0%	99.5%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PLXNA1	100.0%	100.0%	100.0%	100.0%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PLXNA2	100.0%	100.0%	100.0%	99.7%	
PLXND1	100.0%	100.0%	100.0%	99.6%	Congenital heart defects, multiple types, 9, 620294
PMM2	100.0%	100.0%	100.0%	98.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	100.0%	100.0%	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	100.0%	100.0%	100.0%	99.4%	Multiple mitochondrial dysfunctions syndrome 6, 617954

PNKP	100.0%	100.0%	100.0%	99.9%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNP	100.0%	100.0%	100.0%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	100.0%	100.0%	100.0%	99.9%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
PNPT1	100.0%	100.0%	100.0%	99.3%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POGZ	100.0%	100.0%	100.0%	99.3%	White-Sutton syndrome, 616364
POLA1	99.7%	99.4%	98.5%	72.7%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030

POLG	100.0%	100.0%	100.0%	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
POLR1C	83.3%	83.2%	100.0%	99.8%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR2A	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100.0%	100.0%	100.0%	99.5%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100.0%	99.9%	100.0%	99.0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLRMT	100.0%	100.0%	100.0%	99.9%	Combined oxidative phosphorylation deficiency 55, 619743

POMGNT1	100.0%	100.0%	100.0%	99.9%	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
POMGNT2	100.0%	100.0%	100.0%	99.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100.0%	100.0%	100.0%	99.7%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100.0%	100.0%	100.0%	99.8%	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

POMT2	100.0%	100.0%	100.0%	98.8%	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
PORCN	100.0%	99.8%	99.0%	75.5%	Focal dermal hypoplasia, 305600
POU1F1	100.0%	100.0%	100.0%	99.6%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU3F2	100.0%	100.0%	100.0%	98.8%	
POU3F3	99.7%	97.7%	99.7%	84.3%	Snijders Blok-Fisher syndrome, 618604
PPFIBP1	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPIL1	100.0%	100.0%	100.0%	99.6%	Pontocerebellar hypoplasia, type 14, 619301
PPM1D	100.0%	100.0%	100.0%	99.5%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPP1CB	100.0%	100.0%	99.9%	99.0%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	99.9%	99.3%	100.0%	99.4%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100.0%	100.0%	100.0%	99.2%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383
PPP1R3F	100.0%	99.9%	99.3%	80.6%	

PPP2CA	100.0%	100.0%	100.0%	99.3%	Houge-Janssens syndrome 3, 618354
PPP2R1A	93.7%	93.6%	100.0%	99.7%	Houge-Janssens syndrome 2, 616362
PPP2R3C	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PPP2R5B	100.0%	100.0%	100.0%	99.7%	
PPP2R5C	100.0%	100.0%	100.0%	99.4%	
PPP2R5D	100.0%	100.0%	100.0%	99.6%	Houge-Janssens syndrome 1, 616355
PPP3CA	100.0%	99.9%	100.0%	99.4%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PPT1	90.3%	90.3%	100.0%	99.4%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100.0%	100.0%	99.3%	73.8%	Renpenning syndrome, 309500
PRDM13	100.0%	100.0%	100.0%	99.6%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM15	100.0%	99.6%	100.0%	98.0%	
PRICKLE2	100.0%	100.0%	100.0%	99.8%	
PRKACB	99.8%	99.2%	100.0%	99.2%	Cardioacrofacial dysplasia 2, 619143
PRKAR1A	100.0%	100.0%	100.0%	99.6%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960;Adrenocortical tumor, somatic,

PRKAR1B	100.0%	100.0%	100.0%	99.8%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRMT7	100.0%	100.0%	100.0%	99.9%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRODH	100.0%	100.0%	100.0%	99.9%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500
PRPF8	100.0%	100.0%	100.0%	99.5%	Retinitis pigmentosa 13, 600059
PRPS1	100.0%	100.0%	99.0%	75.1%	Arts syndrome, 301835;Phosphoribosylpyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PRR12	100.0%	100.0%	100.0%	99.6%	Neuroocular syndrome, 619539
PRSS12	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder, autosomal recessive 1, 249500
PRUNE1	93.4%	93.1%	100.0%	99.2%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PSAP	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	99.3%	Neu-Laxova syndrome 2, 616038;?Phosphoserine aminotransferase deficiency, 610992

PSMC3	100.0%	100.0%	100.0%	99.7%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354
PSMC5	100.0%	100.0%	100.0%	99.7%	
PSMD12	100.0%	100.0%	100.0%	99.5%	Stankiewicz-Isidor syndrome, 617516
PSPH	100.0%	100.0%	100.0%	98.8%	Phosphoserine phosphatase deficiency, 614023
PTCH1	100.0%	100.0%	100.0%	99.4%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTCHD1	100.0%	99.9%	98.2%	74.8%	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	100.0%	100.0%	100.0%	99.3%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	100.0%	100.0%	100.0%	99.3%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTF1A	100.0%	100.0%	100.0%	98.6%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935
PTPA	100.0%	100.0%	100.0%	99.2%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482
PTPN11	100.0%	100.0%	100.0%	98.9%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785

PTPN23	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTRH2	100.0%	100.0%	100.0%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100.0%	100.0%	100.0%	99.7%	
PTS	100.0%	100.0%	100.0%	99.0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100.0%	100.0%	100.0%	98.8%	Verheij syndrome, 615583
PUM1	100.0%	100.0%	100.0%	99.4%	Spinocerebellar ataxia 47, 617931;Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719
PURA	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100.0%	100.0%	100.0%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051
PUS7	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	100.0%	100.0%	100.0%	100.0%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100.0%	100.0%	100.0%	99.9%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100.0%	100.0%	100.0%	99.7%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760

QDPR	100.0%	100.0%	100.0%	99.0%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100.0%	100.0%	100.0%	99.7%	Ververi-Brady syndrome, 617982
RAB11B	100.0%	100.0%	100.0%	100.0%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100.0%	100.0%	100.0%	97.0%	
RAB18	100.0%	100.0%	100.0%	99.6%	Warburg micro syndrome 3, 614222
RAB23	100.0%	100.0%	100.0%	98.9%	Carpenter syndrome, 201000
RAB27A	100.0%	100.0%	100.0%	100.0%	Griscelli syndrome, type 2, 607624
RAB39B	100.0%	100.0%	99.1%	76.4%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RAB3GAP1	99.0%	99.0%	100.0%	99.0%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	100.0%	100.0%	100.0%	99.0%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RAB5C	100.0%	100.0%	100.0%	99.2%	
RABGAP1	100.0%	100.0%	100.0%	99.5%	
RAC1	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAC3	100.0%	100.0%	100.0%	98.2%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100.0%	100.0%	100.0%	99.4%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376

RAF1	100.0%	100.0%	100.0%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RAI1	100.0%	100.0%	100.0%	99.6%	Smith-Magenis syndrome, 182290
RALA	100.0%	100.0%	100.0%	98.6%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100.0%	99.9%	100.0%	99.3%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797
RARB	100.0%	100.0%	100.0%	99.8%	Microphthalmia, syndromic 12, 615524
RARS1	94.4%	94.3%	100.0%	98.8%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100.0%	100.0%	100.0%	99.5%	Pontocerebellar hypoplasia, type 6, 611523
RBBP8	100.0%	100.0%	100.0%	98.7%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic,
RBFOX1	100.0%	99.7%	100.0%	99.4%	
RBL2	100.0%	100.0%	100.0%	99.6%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM10	100.0%	99.9%	98.8%	79.1%	TARP syndrome, 311900
RBM28	100.0%	100.0%	100.0%	99.3%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	100.0%	100.0%	100.0%	99.4%	Adams-Oliver syndrome 3, 614814
RCBTB1	100.0%	100.0%	100.0%	99.6%	Retinal dystrophy with or without extraocular anomalies, 617175

RECQL4	100.0%	100.0%	100.0%	100.0%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RELN	99.9%	99.7%	100.0%	99.5%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320
RERE	100.0%	99.9%	99.9%	97.7%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	97.7%	97.6%	100.0%	99.1%	
RFT1	100.0%	100.0%	100.0%	99.4%	Congenital disorder of glycosylation, type In, 612015
RFX3	99.5%	98.6%	100.0%	99.5%	
RFX4	100.0%	100.0%	100.0%	99.6%	
RFX7	100.0%	100.0%	99.9%	97.7%	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330
RHEB	100.0%	100.0%	100.0%	99.2%	
RHOBTB2	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 64, 618004
RIC1	100.0%	99.9%	100.0%	99.4%	CATIFA syndrome, 618761
RIMS2	100.0%	99.9%	100.0%	99.2%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIT1	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 8, 615355
RLIM	100.0%	100.0%	99.0%	75.5%	Tonne-Kalscheuer syndrome, 300978
RMND1	100.0%	100.0%	100.0%	99.3%	Combined oxidative phosphorylation deficiency 11, 614922

RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNASEH2A	100.0%	100.0%	100.0%	99.9%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100.0%	98.8%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100.0%	100.0%	100.0%	99.7%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100.0%	99.9%	97.3%	69.4%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100.0%	100.0%	100.0%	99.9%	Tenorio syndrome, 616260
RNF13	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 73, 618379
RNF2	100.0%	100.0%	100.0%	99.7%	Luo-Schoch-Yamamoto syndrome, 619460
RNF220	100.0%	100.0%	100.0%	99.7%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNPC3	100.0%	100.0%	100.0%	98.7%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU12-2P					
RNU4ATAC					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710

ROBO1	100.0%	99.9%	100.0%	99.6%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROGDI	100.0%	100.0%	100.0%	99.8%	Kohlschutter-Tonz syndrome, 226750
ROR2	100.0%	100.0%	100.0%	99.8%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310
RORA	100.0%	100.0%	100.0%	99.0%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORB	100.0%	100.0%	100.0%	99.2%	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357
RPGRIP1L	100.0%	100.0%	100.0%	98.8%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPIA	100.0%	100.0%	100.0%	99.5%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100.0%	99.8%	99.0%	73.2%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPS19	100.0%	100.0%	100.0%	99.5%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	99.9%	99.5%	98.4%	74.0%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600

RRM2B	100.0%	100.0%	100.0%	98.9%	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
RRP7A	100.0%	99.9%	100.0%	98.7%	?Microcephaly 28, primary, autosomal recessive, 619453
RSPRY1	100.0%	100.0%	100.0%	99.2%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	100.0%	99.9%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100.0%	100.0%	100.0%	99.9%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
RTN4IP1	100.0%	100.0%	100.0%	99.4%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RTTN	100.0%	99.9%	100.0%	99.4%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100.0%	100.0%	100.0%	99.5%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUSC2	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder, autosomal recessive 61, 617773

RXYLT1	100.0%	100.0%	100.0%	99.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
SALL1	100.0%	100.0%	100.0%	99.5%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SAMD9	100.0%	100.0%	100.0%	99.0%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMHD1	100.0%	100.0%	100.0%	99.2%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SARS1	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709
SATB1	100.0%	100.0%	100.0%	99.4%	den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100.0%	99.7%	100.0%	99.4%	Glass syndrome, 612313
SBDS	100.0%	100.0%	100.0%	99.0%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SC5D	100.0%	100.0%	100.0%	98.7%	Lathosterolosis, 607330
SCAF4	100.0%	100.0%	100.0%	99.5%	Fliedner-Zweier syndrome, 620511
SCAMP5	100.0%	100.0%	100.0%	99.5%	
SCAPER	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder and retinitis pigmentosa, 618195

SCN1A	100.0%	100.0%	100.0%	99.4%	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
SCN1B	100.0%	100.0%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.4%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
SCN3A	100.0%	100.0%	100.0%	99.0%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN8A	100.0%	100.0%	100.0%	99.3%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558
SCO1	100.0%	100.0%	100.0%	99.8%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100.0%	100.0%	100.0%	99.8%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377

SCUBE3	100.0%	100.0%	100.0%	99.8%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184
SCYL1	100.0%	100.0%	100.0%	99.5%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SDCCAG8	100.0%	100.0%	100.0%	99.0%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SDHA	100.0%	100.0%	100.0%	99.9%	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
SEC31A	100.0%	100.0%	100.0%	99.2%	?Halperin-Birk syndrome, 618651
SEMA3E	100.0%	100.0%	100.0%	99.7%	
SEMA6B	100.0%	100.0%	100.0%	99.8%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	100.0%	100.0%	100.0%	99.5%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100.0%	100.0%	100.0%	99.1%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	100.0%	99.9%	99.7%	93.8%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	100.0%	100.0%	100.0%	98.5%	Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078

SETD1A	100.0%	100.0%	100.0%	99.3%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100.0%	99.8%	99.9%	97.7%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100.0%	100.0%	100.0%	99.1%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SETD5	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal dominant 23, 615761
SFXN4	100.0%	100.0%	100.0%	97.9%	Combined oxidative phosphorylation deficiency 18, 615578
SGPL1	100.0%	100.0%	100.0%	99.5%	RENI syndrome, 617575
SGSH	100.0%	100.0%	100.0%	99.9%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SHANK1	100.0%	100.0%	99.9%	97.6%	
SHANK2	100.0%	100.0%	100.0%	99.6%	{Autism susceptibility 17}, 613436
SHANK3	99.8%	99.3%	99.9%	99.0%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SHH	100.0%	100.0%	100.0%	98.7%	Microphthalmia with coloboma 5, 611638;Schizencephaly, 269160;Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945
SHMT2	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121

SHOC2	100.0%	100.0%	100.0%	98.7%	Noonan syndrome-like with loose anagen hair 1, 607721
SHQ1	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SHROOM4	100.0%	99.9%	98.8%	74.8%	
SIAH1	100.0%	100.0%	100.0%	99.5%	Buratti-Harel syndrome, 619314
SIK1	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 30, 616341
SIL1	100.0%	100.0%	100.0%	99.7%	Marinesco-Sjogren syndrome, 248800
SIN3A	100.0%	100.0%	100.0%	99.5%	Witteveen-Kolk syndrome, 613406
SIN3B	100.0%	100.0%	100.0%	99.7%	
SIX3	100.0%	100.0%	100.0%	98.9%	Schizencephaly, 269160;Holoprosencephaly 2, 157170
SKI	100.0%	99.9%	100.0%	98.6%	Shprintzen-Goldberg syndrome, 182212
SLC12A2	100.0%	100.0%	100.0%	98.9%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC12A5	100.0%	100.0%	100.0%	99.6%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100.0%	100.0%	100.0%	99.5%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068

SLC13A5	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A2	100.0%	99.9%	98.1%	70.9%	Allan-Herndon-Dudley syndrome, 300523
SLC17A5	100.0%	100.0%	100.0%	99.2%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC19A3	99.6%	98.4%	100.0%	99.6%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A1	100.0%	100.0%	100.0%	99.5%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232
SLC1A2	100.0%	99.8%	100.0%	99.4%	Developmental and epileptic encephalopathy 41, 617105
SLC1A4	100.0%	100.0%	100.0%	99.9%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	100.0%	100.0%	100.0%	99.0%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A12	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 39, 612949
SLC25A15	100.0%	100.0%	100.0%	99.9%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99.5%	99.5%	99.3%	97.2%	Fontaine progeroid syndrome, 612289
SLC25A42	100.0%	100.0%	100.0%	99.9%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416

SLC2A1	100.0%	100.0%	100.0%	99.8%	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
SLC30A9	100.0%	100.0%	100.0%	99.5%	Birk-Landau-Perez syndrome, 617595
SLC32A1	100.0%	100.0%	100.0%	99.9%	
SLC33A1	100.0%	100.0%	100.0%	99.4%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC35A1	100.0%	100.0%	100.0%	99.8%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100.0%	100.0%	99.3%	79.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC35A3	97.7%	93.3%	100.0%	97.5%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100.0%	100.0%	100.0%	99.7%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100.0%	100.0%	100.0%	100.0%	Congenital disorder of glycosylation, type IIc, 266265
SLC38A3	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 102, 619881
SLC39A14	93.6%	93.6%	100.0%	99.8%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A8	100.0%	100.0%	100.0%	99.3%	Congenital disorder of glycosylation, type IIn, 616721

SLC45A1	100.0%	100.0%	100.0%	98.5%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC46A1	100.0%	100.0%	100.0%	99.8%	Folate malabsorption, hereditary, 229050
SLC4A4	100.0%	99.7%	100.0%	99.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC5A6	100.0%	100.0%	100.0%	99.8%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903
SLC5A7	100.0%	100.0%	100.0%	99.8%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	100.0%	100.0%	100.0%	99.6%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100.0%	100.0%	100.0%	98.6%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100.0%	100.0%	100.0%	99.8%	Hartnup disorder, 234500
SLC6A3	100.0%	100.0%	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SLC6A8	100.0%	99.6%	98.5%	80.3%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100.0%	100.0%	100.0%	99.9%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100.0%	100.0%	100.0%	99.1%	Lysinuric protein intolerance, 222700
SLC9A6	100.0%	99.9%	98.6%	72.8%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243

SLC9A7	100.0%	99.8%	99.0%	74.8%	Intellectual developmental disorder, X-linked 108, 301024
SLF2	100.0%	100.0%	100.0%	99.2%	Atelis syndrome 1, 620184
SLITRK2	100.0%	100.0%	98.3%	69.6%	Intellectual developmental disorder, X-linked 111, 301107
SMAD4	100.0%	100.0%	100.0%	99.8%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA1	100.0%	99.8%	98.2%	72.4%	
SMARCA2	100.0%	99.8%	100.0%	99.7%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100.0%	100.0%	100.0%	99.8%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCA5	100.0%	100.0%	100.0%	99.0%	
SMARCB1	100.0%	100.0%	100.0%	99.9%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	100.0%	100.0%	100.0%	99.3%	Coffin-Siris syndrome 8, 618362
SMARCD1	100.0%	100.0%	100.0%	98.7%	Coffin-Siris syndrome 11, 618779
SMARCE1	100.0%	100.0%	100.0%	99.5%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938

SMC1A	100.0%	99.8%	98.5%	73.6%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100.0%	100.0%	100.0%	99.1%	Cornelia de Lange syndrome 3, 610759
SMC5	100.0%	100.0%	100.0%	98.4%	Atelis syndrome 2, 620185
SMG8	100.0%	100.0%	100.0%	99.3%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100.0%	100.0%	100.0%	99.8%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995
SMOC1	100.0%	100.0%	100.0%	99.6%	Microphthalmia with limb anomalies, 206920
SMPD1	100.0%	100.0%	100.0%	99.4%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SMPD4	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	100.0%	99.4%	98.6%	76.0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAP25	100.0%	100.0%	100.0%	99.5%	?Myasthenic syndrome, congenital, 18, 616330
SNAP29	100.0%	100.0%	100.0%	99.6%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNAPC4	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515

SNIP1	100.0%	100.0%	100.0%	98.5%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORD118					Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRPB	100.0%	100.0%	100.0%	99.7%	Cerebrocostomandibular syndrome, 117650
SNRPN	100.0%	100.0%	100.0%	99.8%	
SNX14	100.0%	100.0%	100.0%	99.0%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100.0%	100.0%	100.0%	99.5%	
SOBP	100.0%	99.5%	100.0%	98.6%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671
SON	100.0%	100.0%	100.0%	99.7%	ZTTK syndrome, 617140
SOS1	100.0%	100.0%	100.0%	99.0%	Noonan syndrome 4, 610733;?Fibromatosis, gingival, 1, 135300
SOS2	100.0%	100.0%	100.0%	99.1%	Noonan syndrome 9, 616559
SOX10	100.0%	100.0%	100.0%	99.9%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100.0%	100.0%	100.0%	98.5%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX2	100.0%	100.0%	100.0%	99.4%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900

SOX3	100.0%	100.0%	98.2%	74.5%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX4	100.0%	100.0%	100.0%	96.2%	Coffin-Siris syndrome 10, 618506
SOX5	100.0%	99.8%	100.0%	99.1%	Lamb-Shaffer syndrome, 616803
SOX6	99.8%	99.3%	100.0%	99.5%	Tolchin-Le Caignec syndrome, 618971
SPART	100.0%	100.0%	100.0%	99.3%	Troyer syndrome, 275900
SPAST	100.0%	100.0%	100.0%	98.2%	Spastic paraplegia 4, autosomal dominant, 182601
SPATA5	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
SPATA5L1	100.0%	100.0%	100.0%	99.4%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
SPECC1L	100.0%	100.0%	100.0%	99.4%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPEN	100.0%	100.0%	100.0%	99.0%	Radio-Tartaglia syndrome, 619312
SPG11	100.0%	100.0%	100.0%	99.3%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	100.0%	100.0%	100.0%	99.9%	
SPOP	100.0%	100.0%	100.0%	98.9%	Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829

SPR	100.0%	100.0%	100.0%	99.7%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	100.0%	100.0%	100.0%	99.6%	Legius syndrome, 611431
SPRED2	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 14, 619745
SPTAN1	100.0%	100.0%	100.0%	99.6%	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
SPTBN1	100.0%	100.0%	100.0%	99.6%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100.0%	99.8%	100.0%	99.8%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SRCAP	100.0%	100.0%	100.0%	99.6%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRD5A3	100.0%	100.0%	100.0%	99.7%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SRP54	100.0%	100.0%	100.0%	99.7%	Neutropenia, severe congenital, 8, autosomal dominant, 618752

SRPX2	100.0%	99.7%	98.2%	75.7%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM2	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal dominant 72, 620439
SRSF1	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489
SSR4	100.0%	99.9%	98.6%	77.7%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	97.4%	95.3%	100.0%	99.6%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.3%	98.3%	100.0%	99.3%	Salt and pepper developmental regression syndrome, 609056
STAG1	100.0%	100.0%	100.0%	98.5%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	100.0%	100.0%	98.6%	73.3%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STAMBP	100.0%	100.0%	100.0%	99.4%	Microcephaly-capillary malformation syndrome, 614261
STIL	100.0%	100.0%	100.0%	99.3%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	100.0%	100.0%	100.0%	99.7%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100.0%	100.0%	100.0%	99.5%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087

STT3A	100.0%	100.0%	100.0%	99.5%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100.0%	100.0%	100.0%	98.7%	Congenital disorder of glycosylation, type Ix, 615597
STX1A	100.0%	100.0%	100.0%	99.3%	
STX1B	100.0%	100.0%	100.0%	99.3%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	100.0%	99.6%	100.0%	99.4%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073
SUCLG1	100.0%	100.0%	100.0%	98.6%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100.0%	100.0%	100.0%	99.8%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SUMF1	100.0%	100.0%	100.0%	99.8%	Multiple sulfatase deficiency, 272200
SUOX	100.0%	100.0%	100.0%	99.5%	Sulfite oxidase deficiency, 272300
SUPT16H	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480

SURF1	100.0%	100.0%	100.0%	99.4%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	100.0%	100.0%	100.0%	97.8%	Imagawa-Matsumoto syndrome, 618786
SVBP	100.0%	100.0%	100.0%	99.1%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYN1	100.0%	100.0%	98.5%	73.4%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNCRIP	100.0%	100.0%	100.0%	99.0%	
SYNGAP1	100.0%	100.0%	100.0%	98.0%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100.0%	100.0%	100.0%	99.3%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYP	100.0%	99.8%	98.6%	75.5%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	100.0%	99.9%	100.0%	99.0%	Baker-Gordon syndrome, 618218
SZT2	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100.0%	100.0%	100.0%	99.5%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAF1	100.0%	99.9%	98.3%	72.1%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF13	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder, autosomal recessive 60, 617432

TAF1C	100.0%	100.0%	100.0%	99.9%	
TAF2	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF4	89.8%	84.8%	99.0%	85.0%	Intellectual developmental disorder, autosomal dominant 73, 620450
TAF6	100.0%	100.0%	100.0%	99.7%	Alazami-Yuan syndrome, 617126
TAF8	89.3%	89.2%	100.0%	99.8%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TANC2	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100.0%	100.0%	100.0%	99.4%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100.0%	100.0%	100.0%	99.0%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TASP1	99.6%	99.5%	100.0%	99.3%	Suleiman-El-Hattab syndrome, 618950
TAT	100.0%	100.0%	100.0%	99.8%	Tyrosinemia, type II, 276600
TBC1D20	100.0%	100.0%	100.0%	99.2%	Warburg micro syndrome 4, 615663
TBC1D23	100.0%	100.0%	100.0%	99.3%	Pontocerebellar hypoplasia, type 11, 617695

TBC1D24	100.0%	100.0%	100.0%	100.0%	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	99.9%	99.6%	100.0%	99.6%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D7	100.0%	100.0%	100.0%	99.4%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCD	100.0%	100.0%	100.0%	99.8%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100.0%	100.0%	100.0%	99.5%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TBCK	100.0%	100.0%	100.0%	99.5%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342
TBP	100.0%	100.0%	100.0%	99.1%	Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600
TBR1	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder with autism and speech delay, 606053

TBX1	97.7%	95.5%	100.0%	97.6%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TCEAL1	100.0%	100.0%	98.9%	71.2%	Hijazi-Reis syndrome, 301094
TCF20	100.0%	100.0%	100.0%	99.4%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	100.0%	100.0%	100.0%	99.3%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100.0%	100.0%	100.0%	97.3%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	100.0%	100.0%	100.0%	99.7%	Transcobalamin II deficiency, 275350
TCTN2	100.0%	100.0%	100.0%	99.6%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100.0%	100.0%	100.0%	99.4%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDP2	100.0%	100.0%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100.0%	100.0%	100.0%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	100.0%	100.0%	100.0%	99.9%	Intellectual developmental disorder, autosomal recessive 14, 614020
TELO2	100.0%	100.0%	100.0%	99.9%	You-Hoover-Fong syndrome, 616954
TENM3	100.0%	100.0%	100.0%	99.8%	Microphthalmia, syndromic 15, 615145;?Microphthalmia, isolated, with coloboma 9, 615145

TET3	100.0%	100.0%	100.0%	99.7%	Beck-Fahrner syndrome, 618798
TFAP2A	100.0%	100.0%	99.9%	96.2%	Branchiooculofacial syndrome, 113620
TFE3	100.0%	99.7%	99.0%	73.4%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854
TGDS	100.0%	100.0%	100.0%	99.0%	Catel-Manzke syndrome, 616145
TGFBR1	100.0%	100.0%	100.0%	99.4%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGIF1	100.0%	100.0%	100.0%	99.3%	Holoprosencephaly 4, 142946
TH	100.0%	100.0%	100.0%	99.7%	Segawa syndrome, recessive, 605407
THG1L	100.0%	100.0%	100.0%	99.2%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC2	100.0%	100.0%	98.7%	73.4%	Intellectual developmental disorder, X-linked 12, 300957
THOC6	100.0%	100.0%	100.0%	99.9%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100.0%	100.0%	100.0%	99.4%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
THUMPD1	100.0%	99.9%	100.0%	98.2%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989
TIAM1	100.0%	100.0%	100.0%	99.5%	Neurodevelopmental disorder with language delay and seizures, 619908

TIMM50	100.0%	100.0%	100.0%	99.8%	3-methylglutaconic aciduria, type IX, 617698
TIMM8A	100.0%	99.5%	97.2%	69.2%	Mohr-Tranebjaerg syndrome, 304700
TINF2	100.0%	100.0%	100.0%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TKFC	100.0%	100.0%	100.0%	99.7%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	98.1%	98.1%	100.0%	99.8%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 57, 618050
TMCO1	88.0%	87.7%	100.0%	98.8%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM106B	100.0%	100.0%	100.0%	99.6%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100.0%	100.0%	100.0%	99.6%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM147	100.0%	100.0%	100.0%	100.0%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075
TMEM163	100.0%	100.0%	100.0%	99.3%	Leukodystrophy, hypomyelinating, 25, 620243
TMEM165	100.0%	100.0%	100.0%	99.5%	Congenital disorder of glycosylation, type IIk, 614727
TMEM216	100.0%	100.0%	100.0%	99.5%	Joubert syndrome 2, 608091;Meckel syndrome 2, 603194

TMEM218	100.0%	100.0%	100.0%	99.4%	Joubert syndrome 39, 619562
TMEM222	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	100.0%	100.0%	100.0%	99.7%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	100.0%	100.0%	100.0%	99.1%	Joubert syndrome 14, 614424
TMEM240	100.0%	100.0%	99.9%	96.4%	Spinocerebellar ataxia 21, 607454
TMEM63A	100.0%	100.0%	100.0%	99.5%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM63B	100.0%	100.0%	100.0%	99.8%	
TMEM63C	100.0%	100.0%	100.0%	99.5%	Spastic paraplegia 87, autosomal recessive, 619966
TMEM67	99.5%	97.5%	100.0%	98.0%	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	100.0%	100.0%	100.0%	98.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100.0%	100.0%	100.0%	99.8%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMLHE	100.0%	99.4%	99.1%	79.3%	{Autism, susceptibility to, X-linked 6}, 300872
TMTC3	100.0%	99.5%	100.0%	99.0%	Lissencephaly 8, 617255
TMX2	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730

TNIK	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNPO2	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNR	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653
TNRC6B	100.0%	100.0%	100.0%	99.6%	Global developmental delay with speech and behavioral abnormalities, 619243
TOE1	100.0%	100.0%	100.0%	99.5%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100.0%	100.0%	100.0%	99.1%	Joubert syndrome 37, 619185
TOMM70	100.0%	100.0%	100.0%	99.9%	
TOR1A	91.2%	90.6%	100.0%	99.3%	Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100;{Dystonia-1, modifier of},
TP53RK	100.0%	100.0%	100.0%	99.5%	Galloway-Mowat syndrome 4, 617730
TP73	100.0%	100.0%	100.0%	99.9%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPI1	100.0%	100.0%	100.0%	99.7%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	100.0%	100.0%	100.0%	99.7%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100.0%	100.0%	100.0%	99.8%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100.0%	100.0%	100.0%	99.4%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220

TPRKB	82.0%	81.2%	100.0%	99.6%	Galloway-Mowat syndrome 5, 617731
TRA2B	100.0%	100.0%	100.0%	99.5%	
TRAF7	100.0%	100.0%	100.0%	99.9%	Cardiac, facial, and digital anomalies with developmental delay, 618164
TRAIP	100.0%	100.0%	100.0%	99.7%	Seckel syndrome 9, 616777
TRAK1	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100.0%	100.0%	100.0%	99.3%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	100.0%	100.0%	100.0%	99.9%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100.0%	100.0%	100.0%	100.0%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100.0%	100.0%	100.0%	98.4%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100.0%	100.0%	100.0%	98.0%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal recessive 13, 613192
TREX1	100.0%	100.0%	100.0%	100.0%	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

TRIM32	100.0%	100.0%	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM8	100.0%	100.0%	100.0%	99.9%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99.9%	99.7%	100.0%	99.5%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIP12	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIT1	100.0%	100.0%	100.0%	99.7%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100.0%	100.0%	100.0%	99.2%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRNT1	100.0%	100.0%	100.0%	99.3%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	100.0%	100.0%	100.0%	99.5%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224

TRRAP	100.0%	100.0%	100.0%	99.2%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100.0%	100.0%	100.0%	99.4%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100.0%	100.0%	100.0%	99.8%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSEN15	100.0%	100.0%	100.0%	99.7%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100.0%	100.0%	100.0%	99.1%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100.0%	100.0%	100.0%	99.8%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TSFM	94.3%	94.3%	100.0%	99.8%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100.0%	100.0%	100.0%	100.0%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	99.2%	98.5%	99.3%	77.3%	Intellectual developmental disorder, X-linked 58, 300210
TSPOAP1	100.0%	100.0%	100.0%	99.7%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456
TTC19	100.0%	100.0%	100.0%	99.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC37	100.0%	100.0%	100.0%	99.3%	Trichohepatoenteric syndrome 1, 222470

TTC5	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC8	100.0%	99.9%	100.0%	99.5%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTI1	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445
TTI2	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder, autosomal recessive 39, 615541
TUBA1A	100.0%	100.0%	100.0%	99.4%	Lissencephaly 3, 611603
TUBA8	100.0%	100.0%	100.0%	99.7%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840
TUBB	99.6%	98.8%	100.0%	99.8%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	100.0%	100.0%	100.0%	99.8%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100.0%	100.0%	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100.0%	100.0%	100.0%	99.8%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	98.9%	95.9%	100.0%	99.8%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100.0%	100.0%	100.0%	99.6%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	96.7%	96.7%	100.0%	99.8%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737

TUBGCP4	100.0%	100.0%	100.0%	99.3%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100.0%	100.0%	100.0%	99.9%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	100.0%	100.0%	100.0%	98.8%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWNK	100.0%	100.0%	100.0%	99.7%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
U2AF2	100.0%	100.0%	99.9%	96.3%	Developmental delay, dysmorphic facies, and brain anomalies, 620535
UBA5	100.0%	100.0%	100.0%	98.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBAP2L	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494
UBE2A	100.0%	100.0%	99.0%	74.0%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860

UBE3A	100.0%	100.0%	100.0%	99.3%	Angelman syndrome, 105830
UBE3B	100.0%	100.0%	100.0%	99.4%	Kaufman oculocerebrofacial syndrome, 244450
UBE3C	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270
UBE4A	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBR1	98.0%	98.0%	100.0%	99.0%	Johanson-Blizzard syndrome, 243800
UBR7	100.0%	100.0%	100.0%	99.6%	Li-Campeau syndrome, 619189
UBTF	100.0%	100.0%	100.0%	99.1%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100.0%	100.0%	100.0%	99.3%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100.0%	100.0%	100.0%	99.8%	Leukodystrophy, hypomyelinating, 14, 617899
UFSP2	100.0%	100.0%	100.0%	99.5%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
UGDH	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 84, 618792
UGP2	95.8%	94.3%	100.0%	98.6%	Developmental and epileptic encephalopathy 83, 618744
UNC13A	100.0%	100.0%	100.0%	99.7%	
UNC45A	100.0%	100.0%	100.0%	99.6%	Osteootohepatoenteric syndrome, 619377

UNC80	100.0%	100.0%	100.0%	99.4%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	100.0%	100.0%	100.0%	99.4%	Beta-ureidopropionase deficiency, 613161
UPF1	99.6%	98.9%	100.0%	99.7%	
UPF3B	100.0%	99.9%	97.7%	69.0%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UROC1	100.0%	100.0%	100.0%	99.9%	?Urocanase deficiency, 276880
USP27X	100.0%	100.0%	99.2%	76.0%	Intellectual developmental disorder, X-linked 105, 300984
USP7	100.0%	99.9%	100.0%	99.0%	Hao-Fountain syndrome, 616863
USP9X	100.0%	99.8%	98.6%	73.7%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
VAMP1	100.0%	100.0%	100.0%	99.5%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS1	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100.0%	100.0%	100.0%	99.6%	Combined oxidative phosphorylation deficiency 20, 615917

VCP	100.0%	100.0%	100.0%	99.3%	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
VLDLR	100.0%	100.0%	100.0%	99.5%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VPS11	100.0%	100.0%	100.0%	99.8%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	99.6%	99.2%	100.0%	99.4%	Cohen syndrome, 216550
VPS16	100.0%	100.0%	100.0%	99.9%	Dystonia 30, 619291
VPS35L	100.0%	100.0%	100.0%	99.2%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	100.0%	100.0%	99.9%	96.5%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	100.0%	99.8%	100.0%	99.6%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS4A	100.0%	100.0%	100.0%	99.6%	CIMDAG syndrome, 619273
VPS50	100.0%	100.0%	100.0%	99.0%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS53	100.0%	100.0%	100.0%	99.7%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	100.0%	99.8%	100.0%	98.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VWA3B	100.0%	100.0%	100.0%	99.1%	?Spinocerebellar ataxia, autosomal recessive 22, 616948

WAC	100.0%	100.0%	100.0%	98.7%	Desanto-Shinawi syndrome, 616708
WARS1	100.0%	100.0%	100.0%	99.3%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelopmental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WARS2	100.0%	100.0%	100.0%	99.3%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	100.0%	99.9%	100.0%	99.4%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder, autosomal recessive 43, 615817
WDFY3	100.0%	100.0%	100.0%	99.5%	?Microcephaly 18, primary, autosomal dominant, 617520
WDPCP	97.5%	97.3%	100.0%	99.6%	?Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100.0%	99.6%	99.7%	81.8%	
WDR26	100.0%	100.0%	100.0%	98.0%	Skraban-Deardorff syndrome, 617616
WDR37	100.0%	100.0%	100.0%	99.6%	Neurooculocardiogenitourinary syndrome, 618652

WDR4	100.0%	100.0%	100.0%	99.4%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR45	100.0%	100.0%	99.7%	84.2%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	100.0%	100.0%	100.0%	98.6%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR5	100.0%	100.0%	100.0%	99.8%	
WDR62	100.0%	100.0%	100.0%	99.8%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
WDR73	100.0%	100.0%	100.0%	99.8%	Galloway-Mowat syndrome 1, 251300
WDR81	100.0%	100.0%	100.0%	99.9%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967
WFS1	100.0%	100.0%	100.0%	99.9%	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
WIPI2	100.0%	100.0%	100.0%	99.3%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WNK3	100.0%	100.0%	98.4%	72.8%	Prieto syndrome, 309610

WNT1	100.0%	100.0%	100.0%	99.8%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WWOX	100.0%	100.0%	100.0%	99.7%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	100.0%	100.0%	100.0%	99.4%	Xeroderma pigmentosum, group A, 278700
XRCC4	100.0%	100.0%	100.0%	98.4%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100.0%	99.8%	100.0%	98.6%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
YARS1	100.0%	100.0%	100.0%	99.1%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YIF1B	90.0%	90.0%	100.0%	99.4%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100.0%	100.0%	100.0%	99.5%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	100.0%	100.0%	100.0%	99.0%	?Optic atrophy 11, 617302
YWHAE	100.0%	100.0%	100.0%	99.2%	
YWHAG	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 56, 617665
YY1	100.0%	99.9%	100.0%	95.2%	Gabriele-de Vries syndrome, 617557
YY1AP1	100.0%	100.0%	100.0%	99.5%	Grange syndrome, 602531
ZBTB11	100.0%	100.0%	100.0%	99.7%	Intellectual developmental disorder, autosomal recessive 69, 618383

ZBTB16	100.0%	100.0%	100.0%	99.9%	Leukemia, acute promyelocytic, PL2F/RARA type,
ZBTB18	100.0%	100.0%	100.0%	99.2%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100.0%	100.0%	100.0%	99.7%	Primrose syndrome, 259050
ZBTB24	100.0%	100.0%	100.0%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB47	100.0%	100.0%	100.0%	99.3%	
ZBTB7A	100.0%	100.0%	100.0%	100.0%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100.0%	99.9%	97.6%	65.7%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZDHHC9	100.0%	99.9%	98.6%	75.1%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799
ZEB2	96.8%	96.7%	100.0%	99.3%	Mowat-Wilson syndrome, 235730
ZFHX4	99.7%	98.9%	100.0%	98.5%	?Ptosis, congenital, 178300
ZFYVE26	100.0%	100.0%	100.0%	99.8%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	100.0%	100.0%	100.0%	99.8%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100.0%	99.9%	100.0%	98.3%	Holoprosencephaly 5, 609637

ZMIZ1	100.0%	99.9%	100.0%	99.6%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYM2	100.0%	100.0%	100.0%	99.6%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522
ZMYM3	100.0%	99.5%	99.0%	75.7%	Intellectual developmental disorder, X-linked 112, 301111
ZMYND11	100.0%	100.0%	100.0%	99.5%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZMYND8	100.0%	100.0%	100.0%	99.6%	
ZNF142	100.0%	100.0%	100.0%	99.9%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	100.0%	100.0%	100.0%	99.0%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99.4%	99.4%	100.0%	99.2%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100.0%	100.0%	100.0%	99.8%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	100.0%	100.0%	100.0%	99.6%	SIMHA syndrome, 619557
ZNF41	100.0%	100.0%	98.2%	75.6%	
ZNF462	100.0%	100.0%	100.0%	99.5%	Weiss-Kruszka syndrome, 618619
ZNF526	100.0%	100.0%	100.0%	99.9%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF668	100.0%	100.0%	100.0%	100.0%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194
ZNF699	100.0%	100.0%	100.0%	99.0%	DEGCAGS syndrome, 619488

ZNF711	100.0%	100.0%	99.4%	76.7%	Intellectual developmental disorder, X-linked 97, 300803
ZNHIT3	78.2%	76.2%	100.0%	98.8%	PEHO syndrome, 260565
ZSWIM6	97.5%	95.9%	98.0%	93.0%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671

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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 3.7.0.