INTELLECTUAL DISABILITY / DEVELOPMENTAL DELAY PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.0.0 (2012 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100%	100%	100%	99.9%	99.3%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS1	100%	100%	100%	100%	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%	100%	100%	100%	99.8%	Hyperlysinemia, 238700
ABAT	100%	100%	100%	99.9%	99.4%	GABA-transaminase deficiency, 613163
ABCA2	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808

ABCC8	100%	100%	100%	100%	99.4%	Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450
ABCC9	96%	96%	100%	100%	99.7%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABCD1	90.6%	86.6%	98.4%	87.4%	67%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABCD4	100%	99.9%	100%	99.9%	99.2%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ABHD16A	97.5%	97.5%	100%	100%	99%	Spastic paraplegia 86, autosomal recessive, 619735
ABHD5	100%	100%	100%	100%	99.7%	Chanarin-Dorfman syndrome, 275630
ACAD9	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 20, 611126
ACADS	100%	100%	100%	100%	99.7%	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470

ACADSB	100%	100%	100%	100%	99.7%	2-methylbutyrylglycinuria, 610006
ACAT1	100%	100%	100%	100%	99.7%	Alpha-methylacetoacetic aciduria, 203750
ACBD6	79.5%	79.5%	100%	100%	99.7%	Neurodevelopmental disorder with progressive movement abnormalities, 620785
ACER3	96.3%	96.3%	100%	100%	99.8%	?Leukodystrophy, progressive, early childhood-onset, 617762
ACO2	92.3%	88.1%	100%	100%	99.4%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACOX1	97.9%	97.9%	100%	100%	99.5%	Mitchell syndrome, 618960;Peroxisomal acyl-CoA oxidase deficiency, 264470
ACSF3	94.7%	94.7%	100%	99.9%	99.5%	Combined malonic and methylmalonic aciduria, 614265
ACSL4	100%	100%	99.4%	92.1%	73%	Intellectual developmental disorder, X-linked 63, 300387
ACTB	100%	100%	100%	100%	99.8%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470

ACTG1	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ACTL6A	100%	100%	100%	100%	99.9%	
ACTL6B	96.2%	94.7%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACVR1	100%	100%	100%	100%	99.8%	Fibrodysplasia ossificans progressiva, 135100
ACY1	100%	100%	100%	100%	99.6%	Aminoacylase 1 deficiency, 609924
ADAM22	98.7%	98.7%	100%	100%	99.8%	Developmental and epileptic encephalopathy 61, 617933
ADAR	100%	100%	100%	100%	99.7%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADARB1	95.1%	94.7%	100%	100%	99.5%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADAT3	100%	100%	100%	100%	99%	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286

ADCY5	97.5%	97.1%	100%	99.9%	98.8%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADD1	100%	100%	100%	100%	99.6%	{Hypertension, essential, salt-sensitive}, 145500
ADD3	98.8%	97.5%	100%	100%	99.7%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100%	100%	100%	100%	99.4%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854
ADGRL1	100%	100%	100%	99.9%	98.9%	Developmental delay, behavioral abnormalities, and neuropsychiatric disorders, 620065
ADK	88.1%	88%	100%	100%	99.9%	Hypermethioninemia due to adenosine kinase deficiency, 614300
ADNP	100%	100%	100%	100%	99.8%	Helsmoortel-van der Aa syndrome, 615873
ADPRS	100%	100%	100%	99.9%	99.5%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
ADSL	100%	99%	100%	100%	99.5%	Adenylosuccinase deficiency, 103050

AFF2	100%	100%	99%	88.7%	68.7%	Intellectual developmental disorder, X-linked 109, 309548
AFF3	100%	100%	100%	100%	99.4%	KINSSHIP syndrome, 619297
AFF4	100%	100%	100%	100%	99.9%	CHOPS syndrome, 616368
AFG2A	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AFG2B	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 119, 619615;Neurodevelopmental disorder with hearing loss and spasticity, 619616
AFG3L2	95.7%	93.1%	100%	100%	99.7%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGA	91.6%	91.6%	100%	100%	99.7%	Aspartylglucosaminuria, 208400
AGAP1	100%	100%	100%	99.8%	98.3%	
AGMO	100%	100%	100%	100%	100%	
AGO1	99.9%	98.9%	100%	100%	99.3%	Neurodevelopmental disorder with language delay and behavioral abnormalities, with or without seizures, 620292
AGO2	100%	100%	100%	100%	99.3%	Lessel-Kreienkamp syndrome, 619149
AGTPBP1	96.8%	96.5%	100%	100%	99.8%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276

AHCY	100%	100%	100%	100%	99.8%	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752
AHDC1	100%	100%	100%	99.8%	98.3%	Xia-Gibbs syndrome, 615829
AHI1	98.7%	98.7%	100%	99.9%	99.4%	Joubert syndrome 3, 608629
AHSG	100%	100%	100%	100%	99.6%	?Alopecia-intellectual disability syndrome 1, 203650
AIFM1	100%	100%	98.5%	88.1%	70.6%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AIMP1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 3, 260600
AIMP2	100%	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 17, 618006
AJAP1	100%	100%	100%	100%	98%	
AKT3	94.6%	94.6%	100%	100%	99.7%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALDH18A1	100%	100%	100%	100%	99.5%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603

ALDH3A2	93.5%	93.5%	100%	100%	99.8%	Sjogren-Larsson syndrome, 270200
ALDH4A1	98.3%	96.2%	100%	100%	99.5%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	100%	99.5%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	97.2%	97.2%	100%	100%	99.6%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALG1	100%	100%	100%	100%	99.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	91%	91%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ip, 613661
ALG12	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type Ig, 607143
ALG13	100%	100%	99.1%	90.9%	71.6%	Developmental and epileptic encephalopathy 36, 300884
ALG14	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG2	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ii, 607906;Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228

ALG3	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100%	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
ALG8	79.8%	77.9%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ALG9	100%	100%	100%	100%	99.5%	Gillessen-Kaesbach-Nishimura syndrome, 263210;Congenital disorder of glycosylation, type Il, 608776
ALKBH8	96.3%	96.3%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 71, 618504
ALMS1	100%	100%	100%	100%	99.7%	Alstrom syndrome, 203800
ALX3	100%	100%	100%	99.9%	98.4%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	100%	100%	99.7%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMER1	100%	100%	98.7%	88.1%	68.4%	Osteopathia striata with cranial sclerosis, 300373
AMFR	100%	100%	100%	100%	99.5%	Spastic paraplegia 89, autosomal recessive, 620379
AMMECR1	99.9%	98.7%	99.2%	90.4%	69.5%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMOTL1	100%	100%	100%	100%	99.5%	Craniofaciocardiohepatic syndrome, 621192

AMPD2	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
AMT	100%	100%	100%	99.9%	99.2%	Glycine encephalopathy 2, 620398
ANK2	100%	100%	100%	100%	99.8%	Long QT syndrome 4, 600919;Cardiac arrhythmia, ankyrin-B-related, 600919
ANK3	99.7%	99.7%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 37, 615493
ANKH	100%	100%	100%	100%	99.6%	Chondrocalcinosis 2, 118600;Craniometaphyseal dysplasia, 123000
ANKLE2	100%	100%	100%	100%	99.4%	Microcephaly 16, primary, autosomal recessive, 616681
ANKRD11	100%	100%	100%	100%	99.3%	KBG syndrome, 148050
ANKRD17	100%	100%	100%	100%	99.7%	Chopra-Amiel-Gordon syndrome, 619504
ANKS1B	100%	100%	100%	100%	99.8%	
ANO10	94.9%	94.9%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO4	100%	100%	100%	100%	99.8%	
ANTXR1	100%	100%	100%	100%	99.4%	GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
AP1B1	93.8%	92.3%	100%	100%	99.6%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

AP1G1	100%	100%	100%	100%	99.9%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP1S1	100%	100%	100%	100%	99.5%	MEDNIK syndrome, 609313
AP1S2	100%	100%	99.7%	91.3%	73.5%	Pettigrew syndrome, 304340
AP2M1	100%	100%	100%	100%	99.3%	Intellectual developmental disorder 60 with seizures, 618587
AP2S1	82.5%	82.5%	100%	99.9%	97.7%	Hypocalciuric hypercalcemia, type III, 600740
AP3B1	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 2, 608233
AP3B2	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 48, 617276
AP3D1	100%	100%	100%	100%	99.2%	?Hermansky-Pudlak syndrome 10, 617050
AP4B1	100%	100%	100%	100%	99.9%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100%	100%	100%	100%	99.6%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	100%	100%	99.3%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	79.1%	79.1%	100%	100%	99.8%	Spastic paraplegia 52, autosomal recessive, 614067

APC2	100%	100%	100%	99.9%	98.9%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
APTX	100%	100%	100%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARCN1	97.2%	96.1%	100%	100%	99.8%	Short stature-micrognathia syndrome, 617164
ARF1	100%	100%	100%	100%	99.8%	Periventricular nodular heterotopia 8, 618185
ARF3	100%	100%	100%	100%	100%	
ARFGEF1	100%	100%	100%	100%	99.8%	Developmental delay, impaired speech, and behavioral abnormalities, with or without seizures, 619964
ARFGEF2	100%	100%	100%	100%	99.7%	Periventricular heterotopia with microcephaly, 608097
ARG1	93%	93%	100%	100%	99.9%	Argininemia, 207800
ARHGAP31	100%	100%	100%	100%	99.7%	Adams-Oliver syndrome 1, 100300
ARHGAP35	100%	100%	100%	100%	99.4%	
ARHGEF6	93.7%	93.6%	99.2%	89.9%	71.3%	
ARHGEF9	96.9%	96.9%	99.1%	90%	70.2%	Developmental and epileptic encephalopathy 8, 300607
ARID1A	100%	100%	100%	100%	99.1%	Coffin-Siris syndrome 2, 614607
ARID1B	98.7%	98.5%	100%	99.7%	96.8%	Coffin-Siris syndrome 1, 135900
ARID2	100%	100%	100%	100%	99.6%	Coffin-Siris syndrome 6, 617808

ARL13B	93.4%	93.4%	100%	100%	99.7%	Joubert syndrome 8, 612291
ARL6	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARMC9	97.2%	94.8%	100%	100%	99.6%	Joubert syndrome 30, 617622
ARPC4	100%	100%	100%	100%	99.6%	Developmental delay, language impairment, and ocular abnormalities, 620141
ARSA	100%	100%	100%	100%	99.2%	Metachromatic leukodystrophy, 250100
ARSL	100%	100%	98.8%	89%	69.1%	Chondrodysplasia punctata, X-linked recessive, 302950
ARV1	90.8%	86.5%	100%	100%	99.6%	Developmental and epileptic encephalopathy 38, 617020
ARX	99.8%	98.1%	95.5%	78.5%	58.1%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100%	100%	100%	100%	99.8%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000

ASH1L	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 52, 617796
ASL	100%	100%	100%	100%	99%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	100%	99.8%	Asparagine synthetase deficiency, 615574
ASPA	100%	100%	100%	100%	99.7%	Canavan disease, 271900
ASPM	97.9%	97.9%	100%	100%	99.7%	Microcephaly 5, primary, autosomal recessive, 608716
ASS1	92.3%	90.9%	100%	100%	99.4%	Citrullinemia, 215700
ASTN1	96.6%	96.6%	100%	100%	99.5%	
ASXL1	100%	100%	100%	99.9%	99.4%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL2	100%	100%	100%	100%	99.6%	Shashi-Pena syndrome, 617190
ASXL3	100%	100%	100%	100%	99.7%	Bainbridge-Ropers syndrome, 615485
ATAD1	96.8%	96.8%	100%	100%	99.7%	Hyperekplexia 4, 618011
ATAD3A	100%	100%	100%	100%	99%	Harel-Yoon syndrome, 617183;Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810
ATCAY	100%	100%	100%	100%	99.3%	Ataxia, cerebellar, Cayman type, 601238
ATG4D	100%	100%	100%	100%	99.4%	Spermatogenic failure 101, 621269
ATG7	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATIC	100%	100%	100%	100%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688

ATL1	100%	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATN1	98.3%	98.2%	100%	99.7%	98%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATOH1	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 89, 620284
ATP13A2	100%	100%	100%	99.9%	98.9%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A1	99.3%	96.1%	100%	100%	99.6%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481

ATP1A3	100%	100%	100%	99.9%	98.7%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2A2	100%	100%	100%	100%	99.7%	Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236
ATP2B1	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 66, 619910
ATP2B2	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 82, 619804
ATP5F1A	100%	100%	100%	100%	99.8%	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
ATP5PO	100%	100%	100%	100%	99.9%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 7, 620359
ATP6AP1	99.7%	98.1%	98.1%	86.2%	66.8%	Immunodeficiency 47, 300972

ATP6AP2	100%	100%	99.5%	93.5%	75.9%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	93%	92.9%	100%	100%	99.6%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0A2	100%	100%	100%	100%	99.8%	Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200
ATP6V0C	100%	100%	100%	100%	99.8%	Epilepsy, early-onset, 3, with or without developmental delay, 620465
ATP6V1A	100%	100%	100%	100%	99.9%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012
ATP6V1B2	94.3%	94.3%	100%	100%	99.9%	Zimmermann-Laband syndrome 2, 616455;Deafness, congenital, with onychodystrophy, autosomal dominant, 124480
ATP7A	94.9%	94.9%	99.2%	91.2%	72.3%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400

ATP8A2	100%	100%	100%	100%	99.6%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268
ATP9A	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with poor growth and behavioral abnormalities, 620242
ATR	100%	100%	99.8%	99.4%	98.9%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
ATRX	100%	100%	99.1%	90.9%	71.8%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
ATXN2L	100%	100%	100%	99.9%	98.7%	
ATXN7L3	100%	100%	100%	99.9%	99.5%	Harel-Tora neurodevelopmental syndrome, 621377
AUH	100%	100%	100%	100%	99.8%	3-methylglutaconic aciduria, type I, 250950
AUTS2	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 26, 615834
AVPR2	100%	100%	97.3%	85.2%	65.4%	Diabetes insipidus, nephrogenic, 1, 304800;Nephrogenic syndrome of inappropriate antidiuresis, 300539

AXIN1	100%	100%	100%	100%	99.4%	Hepatocellular carcinoma, somatic, 114550;Craniometadiaphyseal osteosclerosis with hip dysplasia, 620558;?Caudal duplication anomaly, 607864
B3GALNT2	92.6%	92.6%	100%	100%	99.8%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181
B3GALT6	100%	98.4%	100%	99.9%	97.7%	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%	100%	100%	100%	99.8%	Peters-plus syndrome, 261540
B4GALNT1	100%	100%	100%	100%	99.4%	Spastic paraplegia 26, autosomal recessive, 609195
B4GALT1	91.8%	88.2%	100%	100%	99.6%	Combined low LDL and fibrinogen, 620364;Congenital disorder of glycosylation, type IId, 607091
B4GALT7	86.8%	86.8%	100%	100%	99.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B4GAT1	100%	100%	100%	100%	98.6%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287

B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	99.8%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BAIAP2	100%	100%	100%	99.9%	99.1%	
BAP1	100%	100%	100%	100%	99.3%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BAZ2B	100%	100%	100%	99.8%	99%	
BBS1	93.7%	93.7%	100%	99.9%	99.1%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.6%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.5%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.6%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
BCAP31	91.5%	85.8%	97.6%	86%	68.8%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCAS3	100%	100%	100%	100%	99.8%	Hengel-Maroofian-Schols syndrome, 619641

BCAT2	100%	100%	100%	100%	99.4%	Hypervalinemia and hyperleucine-isoleucinemia, 618850
BCKDHA	86.3%	86.3%	100%	100%	99.7%	Maple syrup urine disease, type Ia, 248600
BCKDHB	90.8%	88.2%	100%	100%	99.7%	Maple syrup urine disease, type Ib, 620698
BCKDK	100%	100%	100%	100%	98.8%	Branched-chain keto acid dehydrogenase kinase deficiency, 614923
BCL11A	100%	100%	100%	100%	98.7%	Dias-Logan syndrome, 617101
BCL11B	100%	99.8%	100%	99.8%	98.1%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BCOR	100%	100%	98.8%	87.6%	68%	Microphthalmia, syndromic 2, 300166
BCORL1	100%	100%	97.9%	85.5%	64.4%	Shukla-Vernon syndrome, 301029
BCS1L	100%	100%	100%	100%	99.3%	GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BHLHE22	100%	100%	100%	99.6%	96.7%	
BICD2	100%	100%	100%	100%	99.7%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290

BICRA	100%	100%	100%	99.8%	98%	Coffin-Siris syndrome 12, 619325
BLM	95.2%	94.7%	100%	100%	99.8%	Bloom syndrome, 210900
BLOC1S1	100%	100%	100%	100%	99.1%	
BLTP1	100%	100%	100%	100%	99.8%	Alkuraya-Kucinskas syndrome, 617822
BMP4	100%	100%	100%	100%	99.2%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BOLA3	100%	100%	100%	100%	99.8%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BORCS8	84.1%	84.1%	100%	100%	99.1%	Neurodegeneration, infantile-onset, with optic atrophy and brain abnormalities, 620987
BPTF	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies, 617755;{Kaposi sarcoma, susceptibility to}, 148000
BRAF	100%	100%	100%	99.7%	98.3%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980

BRAT1	97.5%	96.8%	100%	99.9%	99.4%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BRD4	100%	100%	100%	99.8%	98.1%	Cornelia de Lange syndrome 6, 620568
BRF1	100%	100%	100%	100%	99%	Cerebellofaciodental syndrome, 616202
BRPF1	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333
BRSK2	100%	100%	100%	100%	99%	
BRWD3	100%	100%	99.1%	90.8%	72%	Intellectual developmental disorder, X-linked 93, 300659
BSCL2	100%	100%	100%	100%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BSN	100%	99.8%	100%	100%	99.1%	
BTD	94.2%	94.2%	100%	100%	99.8%	Biotinidase deficiency, 253260
BUB1	97.3%	97.3%	100%	100%	99.5%	Colorectal cancer with chromosomal instability, somatic, 114500;Microcephaly 30, primary, autosomal recessive, 620183

BUB1B	96.3%	96.3%	100%	100%	99.9%	Colorectal cancer, somatic, 114500;[Premature chromatid separation trait], 176430;Mosaic variegated aneuploidy syndrome 1, 257300
C12orf57	100%	100%	100%	100%	99.4%	Temtamy syndrome, 218340
C2CD3	96%	96%	100%	100%	99.7%	Orofaciodigital syndrome XIV, 615948
C2orf69	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 53, 619423
CA2	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CA5A	84.7%	84.7%	100%	100%	99.3%	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751
CA8	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 34, 613227
CACHD1	100%	100%	100%	100%	99.6%	
CACNA1A	100%	100%	100%	100%	99%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500
CACNA1B	97.2%	97.2%	100%	99.9%	99.2%	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497

CACNA1C	100%	100%	100%	99.9%	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%	100%	100%	100%	99.6%	Primary aldosteronism, seizures, and neurologic abnormalities, 615474;Sinoatrial node dysfunction and deafness, 614896
CACNA1E	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 69, 618285
CACNA1G	100%	100%	100%	100%	99.3%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNA1I	100%	100%	100%	99.9%	98.9%	Neurodevelopmental disorder with speech impairment and with or without seizures, 620114
CACNA2D1	100%	100%	100%	99.9%	99.6%	Developmental and epileptic encephalopathy 110, 620149
CACNA2D2	100%	100%	100%	100%	99.3%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100%	99.1%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 50, 616457

CAMK2A	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 53, 617798;?Intellectual developmental disorder, autosomal recessive 63, 618095
CAMK2B	100%	100%	100%	100%	99.1%	Intellectual developmental disorder, autosomal dominant 54, 617799
CAMK2D	97.2%	97.2%	100%	100%	99.7%	
CAMK2G	99.9%	98.4%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 59, 618522
CAMK4	100%	100%	100%	100%	99.8%	
CAMSAP1	100%	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 12, 620316
CAMTA1	100%	100%	100%	99.9%	99.1%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CANT1	100%	100%	100%	100%	99.5%	Desbuquois dysplasia 1, 251450;Epiphyseal dysplasia, multiple, 7, 617719
CAPN15	100%	100%	100%	99.9%	98.9%	Oculogastrointestinal neurodevelopmental syndrome, 619318
CAPRIN1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782;Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636
CAPZA2	100%	100%	100%	100%	99.9%	

CARS1	100%	100%	100%	100%	99.7%	Microcephaly, developmental delay, and brittle hair syndrome, 618891
CARS2	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 27, 616672
CASK	100%	100%	99.1%	91.2%	72.4%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CASP2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 80, with variant lissencephaly, 620653
CBL	100%	100%	100%	100%	99.5%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBS	88%	85.8%	100%	99.9%	99.2%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CBX1	100%	100%	100%	100%	99.8%	
CC2D1A	100%	100%	100%	99.9%	99.2%	Intellectual developmental disorder, autosomal recessive 3, 608443

CC2D2A	98.2%	98.2%	100%	100%	99.9%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCBE1	100%	100%	100%	100%	99.7%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CCDC174	100%	100%	100%	100%	99.8%	Hypotonia, infantile, with psychomotor retardation, 616816
CCDC186	100%	100%	100%	100%	99.8%	
CCDC22	100%	100%	97.7%	85.6%	66.6%	Ritscher-Schinzel syndrome 2, 300963
CCDC32	100%	100%	100%	99.9%	98.9%	Cardiofacioneurodevelopmental syndrome, 619123
CCDC47	100%	100%	100%	100%	99.9%	Trichohepatoneurodevelopmental syndrome, 618268
CCDC82	100%	100%	100%	100%	99.8%	
CCDC88A	97.4%	97.4%	100%	100%	99.6%	PEHO syndrome-like, 617507
CCDC88C	100%	100%	100%	99.9%	98.7%	?Spinocerebellar ataxia 40, 616053;Hydrocephalus, congenital, 1, 236600
CCND2	100%	100%	100%	100%	99.5%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938
CCNK	100%	99.9%	99.9%	98.2%	93.6%	?Intellectual developmental disorder with hypertelorism and distinctive facies, 618147

CCT3	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with speech or visual impairment and brain hypomyelination, 621034
CDC42	100%	100%	100%	100%	99.8%	Takenouchi-Kosaki syndrome, 616737
CDC42BPB	100%	100%	100%	100%	99.4%	Chilton-Okur-Chung neurodevelopmental syndrome, 619841
CDC6	100%	100%	100%	99.9%	99.6%	Meier-Gorlin syndrome 5, 613805
CDH11	100%	100%	100%	99.9%	99.7%	Teebi hypertelorism syndrome 2, 619736;Elsahy-Waters syndrome, 211380
CDH15	100%	100%	100%	99.9%	98.5%	Intellectual developmental disorder, autosomal dominant 3, 612580
CDH2	100%	100%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDK10	100%	100%	100%	99.9%	99.3%	Al Kaissi syndrome, 617694
CDK13	100%	100%	100%	100%	99.4%	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360
CDK16	100%	100%	97.7%	86.3%	65.5%	
CDK19	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 87, 618916

CDK5RAP2	100%	100%	100%	100%	99.7%	Microcephaly 3, primary, autosomal recessive, 604804
CDK8	92.4%	92.4%	100%	100%	99.8%	Intellectual developmental disorder with hypotonia and behavioral abnormalities, 618748
CDKL5	95.7%	95.7%	98.3%	87.6%	68.2%	Developmental and epileptic encephalopathy 2, 300672
CDKN1C	100%	100%	100%	99.8%	98.6%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CDON	100%	100%	100%	100%	99.9%	Holoprosencephaly 11, 614226
CELF2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 97, 619561
CELF4	100%	100%	100%	100%	98.8%	
CELSR1	100%	100%	100%	99.9%	98.9%	Lymphatic malformation 9, 619319
CELSR3	100%	100%	100%	100%	99.2%	
CENPF	98.6%	98.6%	100%	100%	99.7%	Stromme syndrome, 243605
CEP104	95.3%	95.1%	100%	99.9%	99.3%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988
CEP120	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP135	95.8%	95.8%	100%	100%	99.9%	Microcephaly 8, primary, autosomal recessive, 614673

CEP152	100%	100%	100%	100%	99.8%	Microcephaly 9, primary, autosomal recessive, 614852;Seckel syndrome 5, 613823
CEP290	100%	100%	100%	100%	99.8%	Leber congenital amaurosis 10, 611755;Joubert syndrome 5, 610188;Senior-Loken syndrome 6, 610189;?Bardet-Biedl syndrome 14, 615991;Meckel syndrome 4, 611134
CEP295	100%	100%	100%	100%	99.8%	Seckel syndrome 11, 620767
CEP41	100%	100%	100%	100%	99.9%	Joubert syndrome 15, 614464
CEP55	100%	100%	100%	100%	99.8%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP57	100%	100%	100%	100%	99.8%	Mosaic variegated aneuploidy syndrome 2, 614114
CEP63	92.8%	92.8%	100%	100%	99.6%	?Seckel syndrome 6, 614728
CEP83	100%	100%	100%	100%	99.6%	Nephronophthisis 18, 615862
CEP85L	100%	100%	100%	100%	99.8%	Lissencephaly 10, 618873
CEP89	98.3%	98.3%	100%	100%	99.7%	
CERT1	98.1%	98.1%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies, 616351

CHAMP1	100%	100%	100%	99.9%	99.4%	Neurodevelopmental disorder with hypotonia, impaired language, and dysmorphic features, 616579
CHASERR						Neurodevelopmental disorder with dysmorphic facies, absent speech and ambulation, and brain abnormalities, 621012
CHD1	100%	100%	100%	100%	99.7%	Pilarowski-Bjornsson syndrome, 617682
CHD2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 94, 615369
CHD3	100%	100%	100%	100%	99%	Snijders Blok-Campeau syndrome, 618205
CHD4	100%	100%	100%	100%	99.5%	Sifrim-Hitz-Weiss syndrome, 617159
CHD5	100%	100%	100%	99.9%	99%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHD7	100%	100%	100%	100%	99.7%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CHD8	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with autism and macrocephaly, 615032
CHKA	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with microcephaly, movement abnormalities, and seizures, 620023
CHKB	100%	100%	100%	100%	99.6%	Muscular dystrophy, congenital, megaconial type, 602541

CHMP1A	100%	100%	100%	100%	99.4%	Pontocerebellar hypoplasia, type 8, 614961
CHRM1	100%	100%	100%	100%	99.3%	
CHRNA4	100%	100%	100%	99.6%	96.8%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513
CIAO1	100%	100%	100%	100%	99.4%	Multiple mitochondrial dysfunctions syndrome 10, 620960
CIC	100%	100%	100%	100%	98.8%	Intellectual developmental disorder, autosomal dominant 45, 617600
CIT	95.8%	95.8%	100%	100%	99.4%	Microcephaly 17, primary, autosomal recessive, 617090
CIZ1	100%	100%	100%	99.9%	99%	
CKAP2L	100%	100%	100%	100%	100%	Filippi syndrome, 272440
CLCN3	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with seizures and brain abnormalities, 619517;Neurodevelopmental disorder with hypotonia and brain abnormalities, 619512
CLCN4	100%	100%	99.1%	88.6%	68.9%	Raynaud-Claes syndrome, 300114
CLCN6	97.7%	97.7%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 15, 619173
CLDN11	100%	100%	100%	99.9%	98.7%	Leukodystrophy, hypomyelinating, 22, 619328
CLDN5	100%	100%	100%	99.9%	99.4%	
CLIC2	87.2%	87.2%	99.4%	90.9%	71.7%	
CLIP1	100%	100%	100%	100%	99.6%	

CLN3	93.2%	93.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83%	83%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	98.2%	98.2%	100%	99.9%	99.5%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
CLTC	98%	97.1%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 56, 617854
CNKSR2	100%	100%	99.1%	90.1%	71.4%	Intellectual developmental disorder, X-linked syndromic, Houge type, 301008
CNNM2	100%	100%	100%	100%	99.3%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418

CNOT1	100%	100%	100%	100%	99.8%	Vissers-Bodmer syndrome, 619033;Holoprosencephaly 12, with or without pancreatic agenesis, 618500
CNOT2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with nasal speech, dysmorphic facies, and variable skeletal anomalies, 618608
CNOT3	100%	100%	100%	99.9%	98.6%	Intellectual developmental disorder with speech delay, autism, and dysmorphic facies, 618672
CNOT9	99.7%	97.3%	100%	100%	99.8%	
CNPY3	99.8%	96.8%	100%	99.9%	99%	Developmental and epileptic encephalopathy 60, 617929
CNTNAP1	100%	100%	100%	99.9%	99.1%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
CNTNAP2	100%	100%	100%	100%	99.7%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA8	100%	100%	100%	100%	98.8%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COASY	100%	100%	100%	99.9%	99.2%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COG1	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIg, 611209

COG3	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIbb, 620546
COG4	97%	97%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIj, 613489;Saul-Wilson syndrome, 618150
COG5	96.8%	96.8%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIi, 613612
COG6	100%	100%	100%	100%	100%	Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COG7	95.2%	95.1%	100%	100%	99.6%	Congenital disorder of glycosylation, type IIe, 608779
COG8	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IIh, 611182
COL18A1	99.2%	98.6%	100%	100%	99.2%	Knobloch syndrome, type 1, 267750;Glaucoma, primary closed-angle, 618880
COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780

COL4A2	99%	98.5%	100%	100%	99.4%	Brain small vessel disease 2, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519
COLEC11	100%	100%	100%	100%	99.7%	3MC syndrome 2, 265050
COLGALT1	96.8%	93.3%	100%	99.9%	99.1%	Brain small vessel disease 3, 618360
COPB1	96.3%	96.3%	100%	100%	99.7%	Baralle-Macken syndrome, 619255
COPB2	96.7%	96.7%	100%	100%	99.8%	Osteoporosis, childhood- or juvenile-onset, with developmental delay, 619884;?Microcephaly 19, primary, autosomal recessive, 617800
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	96.5%	96.3%	100%	99.9%	99%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ7	100%	100%	100%	100%	99.8%	Coenzyme Q10 deficiency, primary, 8, 616733;Neuronopathy, distal hereditary motor, autosomal recessive 9, 620402
COQ8A	91%	88.1%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	91.9%	91.9%	100%	99.9%	99.1%	Coenzyme Q10 deficiency, primary, 5, 614654
COX10	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 3, 619046

COX11	100%	100%	100%	99.8%	99%	Mitochondrial complex IV deficiency, nuclear type 23, 620275
COX15	100%	100%	100%	100%	99.4%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
COX16	61.9%	61.9%	100%	99.9%	99.2%	Mitochondrial complex IV deficiency, nuclear type 22, 619355
COX6B1	81.2%	80.9%	100%	100%	99.2%	Mitochondrial complex IV deficiency, nuclear type 7, 619051
CPAP	100%	100%	100%	100%	99.7%	Microcephaly 6, primary, autosomal recessive, 608393;?Seckel syndrome 4, 613676
CPE	97.5%	97.5%	100%	99.9%	99.6%	BDV syndrome, 619326
CPLANE1	100%	100%	100%	100%	99.7%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CPLX1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 63, 617976
CPS1	100%	100%	100%	100%	99.8%	Carbamoylphosphate synthetase I deficiency, 237300
CPSF3	95.9%	95.9%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, hypotonia, nystagmus, and seizures, 619876
CRADD	100%	100%	100%	99.9%	99.6%	Intellectual developmental disorder, autosomal recessive 34, with variant lissencephaly, 614499

CRBN	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 2, 607417
CREBBP	100%	100%	100%	100%	99.4%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CRELD1	100%	100%	100%	100%	99.3%	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217;Jeffries-Lakhani neurodevelopmental syndrome, 620771;{Atrioventricular septal defect, susceptibility to, 2}, 606217
CRIPT	100%	100%	100%	100%	99.7%	Rothmund-Thomson syndrome, type 3, 615789
CRLF1	95.8%	91%	99.7%	98.7%	93.7%	Cold-induced sweating syndrome 1, 272430
CRLS1	92%	86.6%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 57, 620167
CRNKL1	100%	100%	100%	100%	99.4%	
CRPPA	100%	100%	100%	100%	99.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
CSDE1	98.8%	97.9%	100%	100%	99.8%	
CSF1R	97.3%	97.3%	100%	100%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820

CSMD1	100%	100%	100%	99.9%	99.5%	
CSNK1G1	100%	100%	100%	100%	99.7%	
CSNK2A1	94.2%	94.2%	100%	100%	99.6%	Okur-Chung neurodevelopmental syndrome, 617062
CSNK2B	100%	100%	100%	99.9%	99.7%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSPP1	96.9%	96.9%	100%	99.9%	99.4%	Joubert syndrome 21, 615636
CSTB	100%	100%	100%	100%	99.7%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	100%	99.7%	100%	99.9%	99%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	100%	100%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTCF	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTDP1	91.4%	90.5%	100%	100%	99.3%	Congenital cataracts, facial dysmorphism, and neuropathy, 604168
CTNNA2	100%	100%	100%	100%	99.7%	Cortical dysplasia, complex, with other brain malformations 9, 618174

CTNNB1	100%	100%	100%	100%	99.9%	Exudative vitreoretinopathy 7, 617572;Pilomatricoma, somatic, 132600;Colorectal cancer, somatic, 114500;Neurodevelopmental disorder with spastic diplegia and visual defects, 615075;Medulloblastoma, somatic, 155255;Ovarian cancer, somatic, 167000;Hepatocellular carcinoma, somatic, 114550
CTNND1	100%	100%	100%	100%	99.6%	Blepharocheilodontic syndrome 2, 617681
CTNND2	100%	100%	100%	99.8%	98.5%	
CTR9	100%	100%	100%	100%	99.8%	
CTSA	100%	100%	100%	100%	99.6%	Galactosialidosis, 256540
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTTNBP2	100%	100%	100%	100%	99.8%	
CTU2	100%	100%	100%	99.9%	99%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL3	97.8%	97.6%	100%	100%	99.8%	Neurodevelopmental disorder with or without autism or seizures, 619239;Pseudohypoaldosteronism, type IIE, 614496
CUL4B	96.7%	96.7%	99.1%	90.7%	71.5%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354

CUX1	100%	100%	100%	99.9%	98.8%	Neurodevelopmental disorder with developmental delay and with or without motor or speech delay, 618330
CUX2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 67, 618141
CWC27	82.6%	82.6%	100%	100%	99.9%	Retinitis pigmentosa with or without skeletal anomalies, 250410
CWF19L1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYB5R3	95.7%	95.5%	100%	100%	99.7%	Methemoglobinemia, type I, 250800;Methemoglobinemia, type II, 250800
CYFIP2	96.2%	96.2%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 65, 618008
CYP27A1	100%	100%	100%	99.9%	99.2%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100%	100%	100%	99.5%	98.1%	Spastic paraplegia 56, autosomal recessive, 615030
D2HGDH	100%	99.2%	100%	100%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DAG1	100%	100%	100%	100%	99.7%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818
DAGLA	100%	100%	100%	100%	99.2%	Neuroocular syndrome 2, paroxysmal type, 168885

DAP3	93.3%	91.2%	100%	100%	99.6%	Perrault syndrome 7, 621101
DARS1	100%	100%	100%	100%	99.8%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	100%	100%	100%	99.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	100%	100%	100%	100%	99.6%	Maple syrup urine disease, type II, 620699
DCAF17	96.5%	96.5%	100%	100%	99.7%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	99.3%	Mitral valve prolapse 2, 607829;Van Maldergem syndrome 1, 601390
DCPS	100%	100%	100%	100%	99.7%	Al-Raqad syndrome, 616459
DCX	100%	100%	98.6%	89.5%	68.7%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DDB1	92.5%	92.3%	100%	100%	99.6%	White-Kernohan syndrome, 619426
DDC	100%	100%	100%	100%	99.7%	Aromatic L-amino acid decarboxylase deficiency, 608643

DDHD2	100%	100%	100%	100%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033
DDX11	100%	100%	100%	100%	99.7%	Warsaw breakage syndrome, 613398
DDX17	100%	100%	100%	100%	99.6%	
DDX23	100%	100%	100%	100%	99.5%	
DDX39B	100%	100%	100%	100%	99.7%	
DDX3X	99.8%	99.2%	98.3%	89%	71.5%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DDX59	100%	100%	100%	100%	99.7%	Orofaciodigital syndrome V, 174300
DDX6	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with impaired language and dysmorphic facies, 618653
DEAF1	94%	90.5%	100%	100%	98.7%	Vulto-van Silfout-de Vries syndrome, 615828;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures, 617171
DEGS1	100%	100%	100%	99.9%	99.7%	Leukodystrophy, hypomyelinating, 18, 618404
DENND5A	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 49, 617281
DENND5B	96%	96%	100%	100%	99.6%	
DEPDC5	100%	100%	100%	100%	99.6%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504

DHCR24	97.8%	97.8%	100%	100%	99.3%	Desmosterolosis, 602398
DHCR7	96.2%	96.2%	100%	100%	99.4%	Smith-Lemli-Opitz syndrome, 270400
DHDDS	74.6%	74.4%	100%	100%	99.4%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861
DHFR	100%	100%	100%	100%	99.7%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DHPS	98.9%	94.3%	100%	100%	99.2%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHRSX	50%	50%	50%	50%	49.8%	Congenital disorder of glycosylation, type 1DD, 301133
DHTKD1	92.7%	92.7%	100%	100%	99.3%	?Charcot-Marie-Tooth disease, axonal, type 2Q, 615025;Alpha-aminoadipic and alpha-ketoadipic aciduria, 204750
DHX16	100%	100%	100%	100%	99.5%	Neuromuscular disease and ocular or auditory anomalies with or without seizures, 618733
DHX30	100%	100%	100%	99.9%	99.1%	Neurodevelopmental disorder with variable motor and speech impairment, 617804

DHX37	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250
DHX9	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal dominant 75, 620988
DIAPH1	100%	100%	100%	100%	99.6%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DIP2B	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630
DIP2C	100%	100%	100%	99.9%	99.4%	
DIS3L2	100%	100%	100%	100%	99.7%	Perlman syndrome, 267000
DKC1	100%	100%	98.8%	87.4%	68.7%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLAT	100%	100%	100%	100%	99.8%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DLG3	100%	100%	98.9%	88.5%	68.1%	Intellectual developmental disorder, X-linked 90, 300850

DLG4	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 62, 618793
DLL1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizures, 618709
DMD	99.6%	99.6%	99.2%	91.3%	72.5%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200
DMPK	100%	100%	100%	99.9%	99.1%	Myotonic dystrophy 1, 160900
DMXL2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DNAJC12	100%	100%	100%	100%	99.6%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC19	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria, type V, 610198
DNM1	100%	100%	100%	100%	99%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346

DNM1L	100%	100%	100%	100%	99.8%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT3A	100%	100%	100%	99.9%	99.6%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
DNMT3B	100%	100%	100%	100%	99.7%	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860;Facioscapulohumeral muscular dystrophy 4, digenic, 619478
DOCK3	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292
DOCK4	100%	100%	100%	100%	99.7%	
DOCK6	100%	100%	100%	100%	99.3%	Adams-Oliver syndrome 2, 614219
DOCK7	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 23, 615859
DOHH	99.2%	94.2%	100%	99.9%	99%	Neurodevelopmental disorder with microcephaly, cerebral atrophy, and visual impairment, 620066
DOLK	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type Im, 610768

DONSON	100%	100%	100%	100%	99.6%	Microcephaly, short stature, and limb abnormalities, 617604;Microcephaly-micromelia syndrome, 251230
DOT1L	100%	100%	100%	99.9%	99%	Nil-Deshwar neurodevelopmental syndrome, 621265
DPAGT1	100%	100%	100%	100%	99.7%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
DPF2	95.7%	95.7%	100%	100%	99%	Coffin-Siris syndrome 7, 618027
DPH1	100%	100%	100%	99.9%	99.1%	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901
DPH5	75.4%	75.4%	100%	100%	99.9%	Neurodevelopmental disorder with short stature, prominent forehead, and feeding difficulties, 620070
DPM1	99.9%	98.8%	98.4%	95.6%	91.9%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	99.9%	100%	100%	99.9%	Congenital disorder of glycosylation, type Iu, 615042
DPP6	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 33, 616311;{Ventricular fibrillation, paroxysmal familial, 2}, 612956
DPYD	86.9%	86.9%	100%	100%	99.7%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270

DPYS	100%	100%	100%	100%	99.4%	Dihydropyrimidinuria, 222748
DPYSL2	100%	100%	100%	99.9%	99.5%	
DPYSL5	100%	100%	100%	100%	99.7%	Ritscher-Schinzel syndrome 4, 619435
DRG1	92.1%	92.1%	100%	99.9%	99.3%	Tan-Almurshedi syndrome, 620641
DTYMK	95.3%	85.3%	100%	100%	99.3%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DYM	100%	100%	100%	100%	99.9%	Smith-McCort dysplasia, 607326;Dyggve-Melchior-Clausen disease, 223800
DYNC1H1	99.3%	99.3%	100%	100%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563
DYNC1I2	92.4%	92.4%	100%	100%	99.8%	Neurodevelopmental disorder with microcephaly and structural brain anomalies, 618492
DYRK1A	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 7, 614104
EARS2	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 12, 614924
EBF3	100%	100%	100%	100%	99.1%	Hypotonia, ataxia, and delayed development syndrome, 617330

EBP	100%	100%	98.6%	85.4%	65.2%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECHS1	71.5%	69.5%	100%	99.9%	99.2%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EDC3	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal recessive 50, 616460
EDEM3	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIv, 619493
EED	99.1%	99.1%	100%	99.8%	97.9%	Cohen-Gibson syndrome, 617561
EEF1A2	99.2%	95.8%	100%	100%	98.8%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EEF1D	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with thin corpus callosum, hypotonia, and absent language, 621150
EEFSEC	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with progressive spasticity and brain abnormalities, 621102
EFL1	96.3%	96.3%	100%	100%	99.8%	Shwachman-Diamond syndrome 2, 617941
EFNB1	100%	100%	98.8%	86.8%	68.4%	Craniofrontonasal dysplasia, 304110
EFNB2	100%	100%	100%	99.9%	99.5%	
EFTUD2	100%	100%	100%	100%	99.5%	Mandibulofacial dysostosis, Guion-Almeida type, 610536

EHMT1	97.7%	97.7%	100%	100%	99.5%	Kleefstra syndrome 1, 610253
EIF2AK1	96.6%	94.3%	100%	99.9%	99.4%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100%	100%	100%	100%	99.6%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2AK3	100%	100%	100%	100%	99.6%	Wolcott-Rallison syndrome, 226980
EIF2B2	100%	100%	100%	100%	99.1%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312
EIF2B3	100%	100%	100%	100%	99.8%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	100%	99.8%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	100%	99.5%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF2S3	100%	100%	99.4%	90.4%	71.2%	MEHMO syndrome, 300148
EIF3F	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 67, 618295

EIF4A2	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia and speech delay, with or without seizures, 620455
EIF4A3	100%	100%	100%	100%	99%	Robin sequence with cleft mandible and limb anomalies, 268305
EIF5A	100%	100%	100%	100%	99.5%	Faundes-Banka syndrome, 619376
EIPR1	91.2%	91.2%	100%	100%	99.8%	
ELAC2	94.8%	94.8%	100%	100%	99.8%	{Prostate cancer, hereditary, 2, susceptibility to}, 614731;Combined oxidative phosphorylation deficiency 17, 615440
ELFN1	100%	100%	100%	99.9%	98%	Dursun-Ozgul neurodevelopmental syndrome, 621344
ELOVL4	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELP2	96.2%	94.8%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 58, 617270
EMC1	100%	99.1%	100%	100%	99.6%	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875
EMC10	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with dysmorphic facies and variable seizures, 619264
EML1	90.9%	90.9%	100%	99.9%	99.4%	Band heterotopia, 600348

EMX2	100%	100%	100%	99.2%	96%	Schizencephaly, 269160
ENTPD1	100%	100%	100%	100%	99.8%	Spastic paraplegia 64, autosomal recessive, 615683
EP300	100%	100%	100%	100%	99.4%	Menke-Hennekam syndrome 2, 618333;Colorectal cancer, somatic, 114500;Rubinstein-Taybi syndrome 2, 613684
EP400	100%	100%	100%	100%	99.4%	
EPB41L3	100%	99.7%	100%	100%	99.8%	
EPG5	100%	100%	100%	100%	99.7%	Vici syndrome, 242840
EPHA7	100%	100%	100%	100%	99.8%	
EPRS1	96.2%	96.2%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 15, 617951
ERCC1	100%	100%	100%	99.8%	98.8%	Cerebrooculofacioskeletal syndrome 4, 610758
ERCC2	96.4%	96.1%	100%	99.9%	99.1%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100%	100%	100%	100%	99.5%	Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651
ERCC5	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

ERCC6	100%	100%	100%	100%	99.5%	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	95.3%	95.2%	100%	99.7%	98.3%	UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERF	100%	100%	100%	100%	98.8%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERI1	100%	100%	100%	100%	99.7%	Hoxha-Aliu syndrome, 620662;Spondyloepimetaphyseal dysplasia, Guo-Campeau type, 620663
ERLIN2	100%	100%	100%	100%	99.5%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ESAM	100%	97.6%	100%	100%	99.4%	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity, 620371
ESCO2	100%	100%	100%	100%	99.9%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
ETFB	92.9%	92.9%	100%	99.9%	99.2%	Glutaric acidemia IIB, 231680

ETHE1	77.1%	77.1%	100%	100%	99.3%	Ethylmalonic encephalopathy, 602473
EXOC2	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and cerebellar hypoplasia, 619306
EXOC7	98.6%	98.6%	100%	100%	98.9%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOC8	100%	100%	100%	100%	99.5%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXOSC2	91.2%	91.1%	100%	100%	99.6%	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763
EXOSC3	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	99.6%	98.2%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1D, 618065
EXT2	100%	100%	100%	100%	99.7%	Seizures, scoliosis, and macrocephaly syndrome, 616682;Exostoses, multiple, type 2, 133701
EXTL3	100%	100%	100%	100%	99.5%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425

EZH1	100%	100%	100%	100%	99.5%	
EZH2	100%	100%	100%	100%	99.8%	Weaver syndrome, 277590
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAM149B1	100%	100%	100%	100%	99.8%	Joubert syndrome 36, 618763
FAM177A1	86.5%	86.5%	100%	100%	99.7%	Neurodevelopmental disorder with white matter abnormalities and gait disturbance, 621152
FAM20C	99.9%	98%	100%	100%	99.3%	Raine syndrome, 259775
FAM50A	93%	93%	98.1%	85.9%	66.5%	Intellectual developmental disorder, X-linked syndromic, Armfield type, 300261
FAR1	92%	90.9%	100%	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100%	100%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FARSA	100%	100%	100%	100%	99.4%	?Rajab interstitial lung disease with brain calcifications 2, 619013
FARSB	94.1%	94%	100%	100%	99.7%	Rajab interstitial lung disease with brain calcifications 1, 613658
FASTKD2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 44, 618855

FAT4	100%	100%	100%	100%	99.6%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBRSL1	99.9%	98.3%	99.7%	96.3%	88%	
FBXL3	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with short stature, facial anomalies, and speech defects, 606220
FBXL4	95.6%	95.6%	100%	100%	99.8%	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471
FBXO11	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities, 618089
FBXO22	100%	100%	100%	100%	99.8%	Tayoun-Maawali syndrome, 621184
FBXO28	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 100, 619777
FBXO31	100%	100%	100%	100%	99.3%	?Intellectual developmental disorder, autosomal recessive 45, 615979
FBXW11	100%	100%	100%	100%	99.5%	Neurodevelopmental, jaw, eye, and digital syndrome, 618914
FBXW7	100%	100%	100%	100%	99.8%	Developmental delay, hypotonia, and impaired language, 620012
FCSK	100%	100%	100%	100%	99.6%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FDFT1	100%	99.6%	100%	100%	99.5%	Squalene synthase deficiency, 618156

FEM1B	100%	100%	100%	100%	100%	Neurodevelopmental disorder with behavioral, ear, and skeletal abnormalities, 621263
FERRY3	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 66, 618221
FEZF2	100%	100%	100%	99.8%	97.4%	
FGD1	100%	99.9%	98.4%	87.7%	68.8%	Aarskog-Scott syndrome, 305400
FGF12	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 47, 617166
FGF13	100%	99.5%	99.5%	91.1%	72.1%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095
FGF14	100%	100%	100%	100%	99.4%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174
FGFR1	100%	99.5%	100%	100%	99.5%	Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

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

FGFR3	100%	100%	100%	100%	99.4%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FH	100%	100%	100%	100%	99.8%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FIBP	99.7%	96.8%	100%	99.9%	99%	Thauvin-Robinet-Faivre syndrome, 617107
FIG4	98.4%	98.4%	100%	100%	99.7%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FIGN	100%	100%	100%	100%	99.4%	
FILIP1	100%	100%	100%	100%	99.7%	Neuromuscular disorder, congenital, with dysmorphic facies, 620775
FITM2	100%	100%	100%	100%	99.5%	Siddiqi syndrome, 618635

FKRP	100%	100%	100%	100%	99.1%	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	96.1%	94.9%	100%	100%	99.8%	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%	100%	98.4%	87.4%	67%	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	95.6%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelopmental disorder with microcephaly, absent speech, and hypotonia, 621060
FLVCR2	100%	100%	100%	100%	99.8%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
FMN2	100%	100%	100%	99.7%	97.2%	Intellectual developmental disorder, autosomal recessive 47, 616193
FMR1	97%	97%	99.2%	91.8%	72.8%	Fragile X tremor/ataxia syndrome, 300623;Fragile X syndrome, 300624;Premature ovarian failure 1, 311360
FOLR1	100%	100%	100%	100%	99.6%	Neurodegeneration due to cerebral folate transport deficiency, 613068

FOSL2	100%	100%	100%	100%	99.5%	Aplasia cutis-enamel dysplasia syndrome, 620789
FOXG1	100%	100%	100%	99.8%	97.3%	Rett syndrome, congenital variant, 613454
FOXJ1	100%	100%	100%	100%	99%	Ciliary dyskinesia, primary, 43, 618699
FOXP1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with language impairment with or without autistic features, 613670
FOXP2	100%	100%	100%	99.9%	99.5%	Speech-language disorder-1, 602081
FOXP4	100%	100%	100%	100%	99.1%	
FOXRED1	100%	100%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRA10AC1	92.2%	92.2%	100%	99.9%	98.8%	Neurodevelopmental disorder with growth retardation, dysmorphic facies, and corpus callosum abnormalities, 620113
FRAS1	100%	100%	100%	100%	99.6%	Fraser syndrome 1, 219000
FRMD4A	96.6%	96.6%	100%	100%	99.4%	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819
FRMD5	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094
FRMPD4	100%	100%	98.5%	88.5%	68.8%	Intellectual developmental disorder, X-linked 104, 300983

FRRS1L	100%	100%	100%	99.9%	98.1%	Developmental and epileptic encephalopathy 37, 616981
FRYL	100%	100%	100%	99.9%	99.5%	Pan-Chung-Bellen syndrome, 621049
FSD1L	100%	100%	100%	99.9%	99.7%	
FTCD	100%	100%	100%	99.8%	98.4%	Glutamate formiminotransferase deficiency, 229100
FTH1	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517
FTO	94.5%	94.5%	100%	100%	99.8%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
FTSJ1	100%	100%	98.2%	86.4%	65.7%	Intellectual developmental disorder, X-linked 9, 309549
FUCA1	100%	100%	100%	100%	99.8%	Fucosidosis, 230000
FUT8	97.2%	97.2%	100%	100%	100%	Congenital disorder of glycosylation with defective fucosylation 1, 618005
FZR1	99.9%	98%	100%	100%	99.5%	Developmental and epileptic encephalopathy 109, 620145
GABBR1	100%	100%	100%	100%	98.8%	Neurodevelopmental disorder with language delay and variable cognitive abnormalities, 620502

GABBR2	100%	100%	100%	99.9%	99.1%	{Nicotine dependence, protection against}, 188890;{Nicotine dependence, susceptibility to}, 188890;Developmental and epileptic encephalopathy 59, 617904;Neurodevelopmental disorder with poor language and loss of hand skills, 617903
GABRA1	100%	100%	100%	100%	99.5%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRA2	95%	95%	100%	100%	99.9%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780
GABRA3	100%	100%	99.2%	90.1%	69.6%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRA4	100%	100%	100%	100%	99.7%	
GABRA5	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 79, 618559
GABRB1	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 92, 617829

GABRB3	100%	100%	100%	100%	99.4%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113
GABRD	100%	100%	100%	100%	99%	{?Generalized epilepsy with febrile seizures plus, type 5, susceptibility to}, 613060
GABRG2	92.9%	92.9%	100%	100%	99.7%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 89, 619124
GALC	100%	100%	100%	100%	99.7%	Krabbe disease, 245200
GALE	100%	100%	100%	100%	99.8%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GALNT2	100%	100%	100%	100%	99.4%	Congenital disorder of glycosylation, type IIt, 618885
GALT	100%	100%	100%	99.9%	99.2%	Galactosemia, 230400
GAMT	97.8%	94.1%	100%	100%	99.1%	Cerebral creatine deficiency syndrome 2, 612736
GATAD2A	100%	100%	100%	100%	99.3%	
GATAD2B	100%	100%	100%	100%	99.7%	GAND syndrome, 615074

GATM	100%	100%	100%	100%	99.8%	Cerebral creatine deficiency syndrome 3, 612718;Fanconi renotubular syndrome 1, 134600
GBA1	100%	98.8%	100%	99.9%	99.2%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GCH1	100%	100%	100%	100%	99.8%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GCSH	100%	100%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GDI1	100%	100%	98.6%	86.7%	65.9%	Intellectual developmental disorder, X-linked 41, 300849
GEMIN4	100%	100%	100%	99.9%	99.1%	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities, 617913
GEMIN5	100%	100%	100%	99.9%	99.7%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	100%	100%	100%	100%	99.3%	Alexander disease, 203450

GFER	100%	100%	100%	100%	99.3%	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076
GFM1	96.6%	96.6%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 1, 609060
GFM2	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 39, 618397
GIGYF1	100%	100%	100%	99.9%	99.2%	
GJA1	100%	100%	100%	99.9%	99.1%	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%	100%	96.7%	82.1%	62.5%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	100%	99.2%	100%	99.3%	97.1%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GK	96%	96%	99.2%	91.2%	70.7%	Glycerol kinase deficiency, 307030

GLB1	100%	100%	100%	100%	99.8%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLDC	97.3%	97.3%	100%	100%	99.7%	Glycine encephalopathy1, 605899
GLI2	100%	100%	100%	100%	99.3%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GLI3	99.3%	99.3%	100%	100%	99.3%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GLIS3	100%	100%	100%	100%	99.6%	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199
GLRA2	100%	100%	98.7%	88.8%	69.8%	Intellectual developmental disorder, X-linked syndromic, Pilorge type, 301076
GLS	95.8%	95.8%	100%	100%	99.6%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100%	100%	100%	100%	99.7%	Hyperinsulinism-hyperammonemia syndrome, 606762

GLUL	100%	100%	100%	100%	99.6%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806
GLYCTK	100%	100%	100%	100%	99.4%	D-glyceric aciduria, 220120
GM2A	100%	100%	100%	100%	99.2%	GM2-gangliosidosis, AB variant, 272750
GMNN	91.4%	91.4%	100%	100%	99.9%	Meier-Gorlin syndrome 6, 616835
GMPPA	100%	100%	100%	100%	98.9%	Alacrima, achalasia, and impaired intellectual development syndrome, 615510
GMPPB	100%	100%	100%	99.9%	99.3%	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%	100%	100%	100%	100%	Neurodevelopmental disorder with hypotonia, impaired speech, and behavioral abnormalities, 619854
GNAO1	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493

GNAS	100%	99.9%	100%	99.9%	99%	Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080
GNB1	94.3%	94.3%	100%	100%	99.7%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973
GNB2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with hypotonia and dysmorphic facies, 619503;?Sick sinus syndrome 4, 619464
GNB5	100%	100%	100%	99.9%	99.6%	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%	100%	100%	100%	99.7%	Rhizomelic chondrodysplasia punctata, type 2, 222765

GNPTAB	100%	100%	100%	99.9%	99.7%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GNPTG	90.1%	90.1%	100%	99.9%	98.9%	Mucolipidosis III gamma, 252605
GNS	94.3%	94.3%	100%	100%	99.7%	Mucopolysaccharidosis type IIID, 252940
GOLGA2	100%	100%	100%	100%	99.3%	Developmental delay with hypotonia, myopathy, and brain abnormalities, 620240
GON4L	100%	100%	100%	100%	99.5%	Li-Takada-Miyake syndrome, 621212
GOT2	92.8%	92.8%	100%	100%	99.6%	Developmental and epileptic encephalopathy 82, 618721
GPAA1	88.5%	88.5%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPATCH11	100%	100%	100%	100%	99.4%	
GPC3	94%	94%	98.6%	88.5%	69.2%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPC4	100%	100%	98.4%	89.1%	69.8%	Keipert syndrome, 301026
GPHN	95.9%	95.9%	100%	100%	99.9%	Molybdenum cofactor deficiency C, 615501
GPRC5B	100%	100%	100%	100%	99.8%	Megalencephalic leukoencephalopathy with subcortical cysts 3, 620447
GPSM2	95.5%	95.5%	100%	100%	99.9%	Chudley-McCullough syndrome, 604213
GPT2	92.9%	92.9%	100%	100%	99.4%	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281

GRIA1	100%	100%	100%	100%	99.5%	?Intellectual developmental disorder, autosomal recessive 76, 619931;Intellectual developmental disorder, autosomal dominant 67, 619927
GRIA2	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with language impairment and behavioral abnormalities, 618917
GRIA3	100%	100%	99%	88.9%	68.7%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699
GRIA4	100%	100%	100%	99.7%	99.1%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	95.3%	95.3%	100%	99.6%	98.8%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092
GRIN1	100%	100%	100%	100%	98.9%	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	100%	100%	100%	100%	99.7%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	100%	99.8%	100%	99.8%	97.6%	Developmental and epileptic encephalopathy 46, 617162
GRIP1	100%	100%	100%	100%	99.6%	Fraser syndrome 3, 617667
GRM1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRM7	100%	100%	100%	99.9%	98.6%	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922
GRN	100%	100%	100%	99.9%	99.2%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
GSE1	100%	100%	100%	99.9%	98.8%	
GSPT2	100%	100%	99%	91.5%	73.9%	
GSS	100%	100%	100%	100%	99.3%	Anemia, congenital, nonspherocytic hemolytic, 6, glutatione synthetase deficient, 231900;Glutathione synthetase deficiency, 266130

GSX2	100%	100%	100%	99.7%	98.5%	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646
GTF2E2	78.7%	78.7%	100%	100%	99.4%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	44.2%	44.2%	100%	100%	99.7%	Trichothiodystrophy 3, photosensitive, 616395
GTF3C3	99.7%	98.9%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies, brain anomalies, and seizures, 621201
GTF3C5	100%	100%	100%	100%	99.1%	
GTPBP1	100%	99.2%	100%	100%	99.3%	Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1, 620888
GTPBP2	100%	100%	100%	100%	99.5%	Jaberi-Elahi syndrome, 617988
GTPBP3	100%	100%	100%	99.9%	98.8%	Combined oxidative phosphorylation deficiency 23, 616198
GUSB	98.4%	98.4%	100%	100%	99.3%	Mucopolysaccharidosis VII, 253220
H1-4	100%	100%	100%	100%	99.4%	Rahman syndrome, 617537
H3-3A	100%	100%	100%	99.9%	99.5%	Bryant-Li-Bhoj neurodevelopmental syndrome 1, 619720
H3-3B	100%	100%	100%	100%	98.3%	Bryant-Li-Bhoj neurodevelopmental syndrome 2, 619721
H4C3	100%	100%	100%	100%	99.4%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 1, 619758

H4C5	100%	100%	100%	100%	99.8%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 3, 619950
H4C9	100%	100%	100%	99.7%	99.2%	Tessadori-Bicknell-van Haaften neurodevelopmental syndrome 4, 619951
HAAO	100%	99.7%	100%	99.9%	99.6%	Vertebral, cardiac, renal, and limb defects syndrome 1, 617660
HACE1	95.9%	95.9%	100%	99.8%	99.2%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	87.7%	87.7%	100%	100%	99.6%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HADHA	96.4%	95.9%	100%	100%	99.6%	HELLP syndrome, maternal, of pregnancy, 609016;LCHAD deficiency, 609016;Mitochondrial trifunctional protein deficiency 1, 609015;Fatty liver, acute, of pregnancy, 609016
HADHB	94.6%	94.6%	100%	100%	99.6%	Mitochondrial trifunctional protein deficiency 2, 620300
HAX1	100%	100%	100%	100%	99.5%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HCCS	100%	100%	99.3%	90.8%	71.5%	Linear skin defects with multiple congenital anomalies 1, 309801
HCFC1	100%	100%	98.5%	86.6%	66.1%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541

HCN1	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HCN2	95.2%	93.2%	99.6%	97.4%	93%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HDAC3	100%	100%	100%	100%	99.6%	
HDAC4	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with central hypotonia and dysmorphic facies, 619797
HDAC6	95%	94.8%	98.6%	87.3%	67.9%	?Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863
HDAC8	94.6%	94.6%	98.9%	89.4%	69.7%	Cornelia de Lange syndrome 5, 300882
HEATR3	100%	100%	100%	99.9%	99.8%	Diamond-Blackfan anemia 21, 620072
HEATR5B	100%	100%	100%	100%	99.8%	
HECTD1	98.5%	98.5%	100%	100%	99.8%	
HECTD4	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250
HECW2	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268

HEPACAM	100%	100%	100%	99.9%	99.2%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HERC1	100%	100%	100%	100%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
HERC2	100%	100%	100%	100%	99.4%	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%	100%	100%	100%	99.9%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HEXA	100%	100%	100%	99.9%	99.4%	[Hex A pseudodeficiency], 272800;GM2-gangliosidosis, several forms, 272800;Tay-Sachs disease, 272800
HEXB	100%	100%	100%	100%	99.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HGSNAT	83.5%	83.5%	100%	100%	99.9%	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930;Retinitis pigmentosa 73, 616544

HIBCH	100%	100%	100%	100%	99.8%	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620
HID1	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 105 with hypopituitarism, 619983
HIKESHI	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 13, 616881
HIVEP2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 43, 616977
HK1	98.8%	96.8%	100%	100%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelop mental disorder with visual defects and brain anomalies, 618547
HLCS	100%	100%	100%	99.9%	99.3%	Holocarboxylase synthetase deficiency, 253270
HMGB1	100%	100%	100%	100%	99.6%	
HMGCL	76.2%	76.2%	100%	100%	99.4%	HMG-CoA lyase deficiency, 246450
HNMT	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 51, 616739;{Asthma, susceptibility to}, 600807
HNRNPC	87.6%	87.6%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 74, 620688

HNRNPD	100%	100%	100%	100%	99.6%	
HNRNPH1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with craniofacial dysmorphism and skeletal defects, 620083
HNRNPH2	100%	100%	99.4%	89.9%	70.4%	Intellectual developmental disorder, X-linked syndromic, Bain type, 300986
HNRNPK	100%	99%	100%	100%	99.7%	Au-Kline syndrome, 616580
HNRNPR	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HNRNPU	98.6%	98.5%	100%	100%	99.3%	Developmental and epileptic encephalopathy 54, 617391
HOXA1	100%	100%	100%	100%	99.3%	Bosley-Salih-Alorainy syndrome, 601536;Athabaskan brainstem dysgenesis syndrome, 601536
HPD	95.7%	91.5%	100%	99.9%	99.1%	Hawkinsinuria, 140350;Tyrosinemia, type III, 276710
HPDL	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	90.5%	88%	99.3%	91.6%	73.1%	Hyperuricemia, HRPT-related, 300323;Lesch-Nyhan syndrome, 300322

HRAS	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	99.8%	Neurofacioskeletal syndrome with or without renal agenesis, 619194
HS6ST2	100%	100%	98.8%	88.2%	68.3%	?Paganini-Miozzo syndrome, 301025
HSD17B10	100%	100%	98.9%	87.7%	67.3%	HSD10 mitochondrial disease, 300438
HSD17B4	100%	100%	100%	100%	99.8%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSPA9	100%	100%	100%	99.9%	99.6%	Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170
HSPD1	100%	99.5%	100%	100%	99.6%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HTRA2	100%	100%	100%	100%	99.5%	{Parkinson disease 13}, 610297;3-methylglutaconic aciduria, type VIII, 617248

HUWE1	100%	100%	98.8%	88.8%	69.5%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
HYCC1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 5, 610532
HYLS1	100%	100%	100%	100%	99.9%	Hydrolethalus syndrome, 236680
IARS1	100%	100%	100%	100%	99.5%	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093
IARS2	100%	99.9%	100%	100%	99.6%	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007
IBA57	100%	100%	100%	99.8%	98.7%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IDS	100%	100%	99.1%	90%	71.4%	Mucopolysaccharidosis II, 309900
IDUA	96%	96%	100%	99.9%	98.9%	Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IER3IP1	93.3%	77.5%	100%	100%	99.5%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100%	100%	100%	100%	99.9%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250

IFT140	100%	100%	100%	100%	99.5%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	98.5%	100%	100%	99.8%	Bardet-Biedl syndrome 19, 615996
IFT74	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT81	92.5%	92.5%	100%	100%	99.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
IGBP1	100%	100%	99.2%	90.6%	70%	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472
IGF1	100%	100%	100%	100%	99.8%	Insulin-like growth factor I deficiency, 608747
IGF1R	100%	100%	100%	100%	99.5%	Insulin-like growth factor I, resistance to, 270450

IKBKG	94.5%	93.7%	98.6%	90.1%	72.3%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IKZF2	100%	100%	100%	100%	99.8%	Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233
IL1RAPL1	100%	100%	98.8%	88.3%	67.4%	Intellectual developmental disorder, X-linked 21, 300143
IMPA1	81.6%	81.6%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 59, 617323
INPP4A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with growth impairment, quadriparesis, and poor or absent speech, 621354
INPP5E	100%	100%	100%	99.8%	98.6%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INPP5K	100%	99.3%	100%	100%	98.8%	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404

INTS1	99.7%	98.9%	100%	100%	99.4%	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571
INTS11	100%	100%	100%	100%	98.7%	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428
INTS13	100%	100%	100%	100%	99.8%	
INTS6	100%	100%	100%	100%	99.7%	
IPO8	97%	97%	100%	100%	99.7%	VISS syndrome, 619472
IQSEC1	100%	100%	100%	99.8%	98.3%	Intellectual developmental disorder with short stature and behavioral abnormalities, 618687
IQSEC2	99.3%	98.1%	97%	84.5%	65.1%	Intellectual developmental disorder, X-linked 1, 309530
IREB2	98.6%	98.6%	100%	100%	99.8%	Neurodegeneration, early-onset, with choreoathetoid movements and microcytic anemia, 618451
IRF2BPL	100%	100%	99.9%	99%	93.9%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
IRX5	100%	100%	100%	99.9%	97.7%	Hamamy syndrome, 611174
ISCA2	100%	100%	100%	99.8%	99.1%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITGA7	100%	100%	100%	100%	99.5%	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204

ITGAV	99.5%	99.5%	100%	100%	99.7%	Immune dysregulation, neurodevelopmental defects, and colitis, 621375
ITPA	76.9%	76.9%	100%	100%	99.7%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
ITPR1	100%	100%	100%	100%	99.7%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
ITSN1	100%	100%	100%	100%	99.8%	
IVD	90.5%	90%	100%	100%	99.7%	Isovaleric acidemia, 243500
JAG1	100%	100%	100%	100%	99.7%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
JAG2	100%	100%	100%	99.8%	98.7%	Muscular dystrophy, limb-girdle, autosomal recessive 27, 619566
JAM3	99.3%	95.2%	100%	100%	99.6%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
JARID2	100%	100%	100%	99.9%	99.2%	Developmental delay with variable intellectual disability and dysmorphic facies, 620098
JKAMP	100%	100%	100%	100%	99.6%	
JMJD1C	100%	100%	100%	100%	99.8%	

KANK1	98.1%	98.1%	100%	100%	99.5%	Cerebral palsy, spastic quadriplegic, 2, 612900
KANSL1	98.1%	98.1%	100%	100%	99.8%	Koolen-De Vries syndrome, 610443
KARS1	94.3%	94.3%	100%	100%	99.6%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KAT5	100%	100%	100%	99.9%	99.2%	Neurodevelopmental disorder with dysmorphic facies, sleep disturbance, and brain abnormalities, 619103
KAT6A	100%	100%	100%	100%	99.5%	Arboleda-Tham syndrome, 616268
KAT6B	100%	100%	100%	100%	99.7%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KAT8	100%	100%	100%	100%	99.5%	Li-Ghorgani-Weisz-Hubshman syndrome, 618974
KATNB1	100%	100%	100%	100%	99.5%	Lissencephaly 6, with microcephaly, 616212
KATNIP	100%	100%	100%	100%	99.5%	Joubert syndrome 26, 616784
KBTBD2	100%	100%	100%	100%	99.8%	
KCNA2	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100%	100%	100%	99.9%	98.8%	

KCNA4	100%	100%	100%	100%	99.4%	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284
KCNB1	100%	100%	100%	99.9%	99.5%	Developmental and epileptic encephalopathy 26, 616056
KCNB2	100%	100%	100%	100%	99.8%	
KCNC1	100%	100%	100%	99.9%	99.1%	Epilepsy, progressive myoclonic 7, 616187
KCNC2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 103, 619913
KCNC3	100%	99.2%	99.9%	98.7%	93.7%	Spinocerebellar ataxia 13, 605259
KCND1	100%	100%	98.3%	85.2%	65.4%	
KCND2	100%	100%	100%	100%	99.3%	
KCND3	100%	100%	100%	100%	98.8%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNH1	98.6%	98.6%	100%	100%	99.7%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNH5	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 112, 620537
KCNJ10	100%	100%	100%	99.9%	99.6%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780

KCNJ11	100%	100%	100%	100%	99.5%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNJ6	100%	100%	100%	100%	99.9%	Keppen-Lubinsky syndrome, 614098
KCNK3	100%	100%	100%	99.9%	98.9%	Pulmonary hypertension, primary, 4, 615344
KCNK4	99.4%	99.4%	100%	100%	98.9%	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome, 618381
KCNK9	100%	100%	100%	100%	99.3%	Birk-Barel syndrome, 612292
KCNMA1	100%	100%	100%	99.9%	99.5%	{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	100%	100%	100%	99.9%	99%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725

KCNN3	100%	100%	100%	99.9%	99.2%	Zimmermann-Laband syndrome 3, 618658
KCNQ2	94.4%	94.1%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200
KCNQ3	100%	100%	100%	99.9%	99.3%	Seizures, benign neonatal, 2, 121201
KCNQ5	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 46, 617601
KCNT1	100%	100%	100%	100%	99%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 57, 617771
KCTD3	100%	100%	100%	99.9%	99.7%	
KCTD7	100%	100%	100%	100%	99%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM1A	93.5%	93.5%	100%	100%	99.6%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990
KDM2A	100%	100%	100%	99.9%	99.5%	
KDM2B	96.1%	94.2%	100%	99.9%	99%	
KDM3B	100%	100%	100%	100%	99.5%	Diets-Jongmans syndrome, 618846

KDM4B	100%	100%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 65, 619320
KDM5A	100%	100%	100%	100%	99.7%	El Hayek-Chahrour neurodevelopmental syndrome, 620820
KDM5B	97.9%	97.5%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 65, 618109
KDM5C	98.1%	97.8%	98.5%	88%	68.3%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6A	100%	100%	98.7%	90.3%	71.8%	Kabuki syndrome 2, 300867
KDM6B	100%	100%	100%	99.9%	98.7%	Stolerman neurodevelopmental syndrome, 618505
KIAA0586	95.6%	95.6%	100%	100%	99.8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KICS2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 83, 621100
KIDINS220	100%	100%	100%	100%	99.7%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501

KIF11	100%	100%	100%	100%	99.5%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KIF14	97.6%	97.6%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF1A	100%	100%	100%	100%	99.5%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF21B	100%	100%	100%	100%	99.3%	
KIF26A	97.7%	97.6%	100%	100%	99.4%	Cortical dysplasia, complex, with other brain malformations 11, 620156
KIF2A	99.9%	98.8%	100%	100%	99.8%	Cortical dysplasia, complex, with other brain malformations 3, 615411
KIF3B	100%	100%	100%	100%	99.5%	Retinitis pigmentosa 89, 618955
KIF4A	96.1%	96.1%	98.9%	89.6%	70.4%	Taurodontism, microdontia, and dens invaginatus, 313490;Intellectual developmental disorder, X-linked 100, 300923

KIF5A	100%	100%	100%	100%	99.4%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
KIF5B	99.3%	97.2%	100%	100%	99.9%	
KIF5C	100%	100%	100%	100%	99.6%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KIF7	100%	100%	100%	100%	99.1%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	95.9%	95.9%	100%	99.9%	99.3%	Goldberg-Shprintzen megacolon syndrome, 609460
KIRREL3	100%	100%	100%	99.9%	98.8%	
KLF7	100%	100%	100%	99.9%	99.1%	
KLHL13	100%	100%	99.2%	90.8%	71.3%	
KLHL15	89.1%	89.1%	99.5%	92.6%	72.3%	Intellectual developmental disorder, X-linked 103, 300982
KLHL20	93.8%	93.7%	100%	100%	99.7%	
KLHL7	100%	100%	100%	100%	99.8%	Retinitis pigmentosa 42, 612943;PERCHING syndrome, 617055
KMT2A	99.3%	99.2%	100%	100%	99.7%	Wiedemann-Steiner syndrome, 605130

KMT2B	100%	99.6%	100%	99.9%	98.8%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KMT2C	100%	100%	100%	100%	99.6%	Kleefstra syndrome 2, 617768
KMT2D	100%	100%	100%	100%	99.1%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KMT2E	100%	100%	100%	100%	99.6%	O'Donnell-Luria-Rodan syndrome, 618512
KMT5B	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 51, 617788
KNL1	98.8%	98.7%	100%	100%	99.8%	Microcephaly 4, primary, autosomal recessive, 604321
KPTN	100%	100%	100%	99.8%	98.7%	Intellectual developmental disorder, autosomal recessive 41, 615637

KRAS	100%	100%	100%	100%	99.7%	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%	100%	98%	85.7%	66.1%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
L2HGDH	92.8%	92.8%	100%	100%	99.7%	L-2-hydroxyglutaric aciduria, 236792
LAGE3	100%	100%	99.8%	91.4%	70.7%	Galloway-Mowat syndrome 2, X-linked, 301006
LAMA1	98.9%	98.9%	100%	100%	99.7%	Poretti-Boltshauser syndrome, 615960

LAMA2	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138;Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855
LAMB1	100%	100%	100%	100%	99.7%	Lissencephaly 5, 615191
LAMB2	100%	100%	100%	100%	99.4%	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199;Pierson syndrome, 609049
LAMC3	100%	100%	100%	100%	99.5%	Cortical malformations, occipital, 614115
LAMP2	71%	70.7%	98.9%	90.1%	70.6%	Danon disease, 300257
LARGE1	97.3%	96.9%	100%	100%	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
LARP1	100%	100%	100%	99.9%	99.3%	
LARP7	100%	100%	100%	100%	99.9%	Alazami syndrome, 615071
LARS1	94.3%	94.3%	100%	99.9%	99.6%	?Infantile liver failure syndrome 1, 615438
LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LAS1L	96%	96%	98.1%	86.7%	68%	Wilson-Turner syndrome, 309585
LDB1	100%	100%	100%	100%	99.5%	

LETM1	93%	90.2%	100%	100%	99.3%	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089
LGI1	100%	100%	100%	100%	99.9%	Epilepsy, familial temporal lobe, 1, 600512
LGI3	100%	100%	100%	99.9%	99.4%	Intellectual developmental disorder with muscle tone abnormalities and distal skeletal defects, 620007
LHX2	100%	100%	100%	100%	99.2%	
LIAS	100%	100%	100%	100%	99.8%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIG4	100%	100%	100%	100%	99.9%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LINGO1	100%	100%	100%	99.8%	97.6%	Intellectual developmental disorder, autosomal recessive 64, 618103
LINS1	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 27, 614340
LIPT2	100%	100%	100%	100%	99.6%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
LMAN2L	100%	100%	100%	100%	99.8%	?Intellectual developmental disorder, autosomal dominant 69, 617863;?Intellectual developmental disorder, autosomal recessive 52, 616887

LMBRD1	86.4%	86.4%	100%	100%	99.9%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMBRD2	100%	100%	100%	100%	99.8%	Developmental delay with variable neurologic and brain abnormalities, 619694
LMNB1	100%	100%	100%	100%	99.8%	Microcephaly 26, primary, autosomal dominant, 619179
LMNB2	92.9%	92.9%	100%	99.7%	97.6%	Microcephaly 27, primary, autosomal dominant, 619180;?Epilepsy, progressive myoclonic, 9, 616540;{Lipodystrophy, partial, acquired, susceptibility to}, 608709
LNPK	85.8%	85.8%	100%	100%	99.7%	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090
LONP1	99.7%	97.7%	100%	100%	99.4%	CODAS syndrome, 600373
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448
LRPPRC	96.8%	96.7%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111
LRRC7	100%	100%	100%	100%	99.7%	
LSM1	100%	100%	100%	100%	99.1%	FICUS syndrome, 621193
LSS	100%	100%	100%	100%	99.8%	Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840

LTBP1	100%	100%	100%	100%	99.5%	Cutis laxa, autosomal recessive, type IIE, 619451
LYRM7	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 8, 615838
LYST	99.5%	99.5%	100%	100%	99.8%	Chediak-Higashi syndrome, 214500
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994
LZTR1	100%	100%	100%	100%	99.4%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAB21L1	100%	100%	100%	100%	99.2%	Cerebellar, ocular, craniofacial, and genital syndrome, 618479
MAB21L2	100%	100%	100%	99.9%	99%	Microphthalmia/colobo ma and skeletal dysplasia syndrome, 615877
MACF1	100%	100%	100%	100%	99.6%	Lissencephaly 9 with complex brainstem malformation, 618325
MADD	98.7%	98.7%	100%	100%	99.3%	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia, 619005;DEEAH syndrome, 619004
MAF	94.9%	91.7%	100%	99.4%	95.6%	Cataract 21, multiple types, 610202;Ayme-Gripp syndrome, 601088
MAG	100%	100%	100%	100%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MAGEL2	100%	100%	100%	100%	99.4%	Schaaf-Yang syndrome, 615547

MAN1B1	100%	99%	100%	100%	99.4%	Rafiq syndrome, 614202
MAN2A2	100%	100%	100%	99.9%	99.4%	
MAN2B1	100%	100%	100%	100%	99%	Mannosidosis, alpha-, types I and II, 248500
MAN2C1	98.4%	98.4%	100%	100%	99.4%	Congenital disorder of deglycosylation 2, 619775
MANBA	100%	99.6%	100%	100%	99.7%	Mannosidosis, beta, 248510
MAOA	100%	100%	98.8%	90.6%	71.8%	Brunner syndrome, 300615
MAP1B	100%	100%	100%	100%	99.6%	?Deafness, autosomal dominant 83, 619808;Periventricular nodular heterotopia 9, 618918
MAP2K1	95.8%	95.8%	100%	100%	99.7%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	92.4%	92.4%	100%	100%	99.1%	Cardiofaciocutaneous syndrome 4, 615280
MAP2K4	100%	100%	100%	100%	99.7%	
MAP4K4	100%	100%	100%	100%	99.6%	
MAPK1	100%	100%	100%	100%	99.7%	Noonan syndrome 13, 619087
MAPK8IP3	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAPKAPK5	100%	100%	100%	100%	99.7%	Neurocardiofaciodigital syndrome, 619869
MAPRE2	100%	100%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 2, 616734

MARK2	100%	100%	100%	99.9%	99.1%	Intellectual developmental disorder, autosomal dominant 76, 621285
MARS1	90.4%	88.8%	100%	100%	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%	100%	100%	100%	99.8%	3MC syndrome 1, 257920
MAST1	100%	100%	100%	100%	99.2%	Mega-corpus-callosum syndrome with cerebellar hypoplasia and cortical malformations, 618273
MAST3	100%	100%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 108, 620115
MAST4	100%	100%	100%	100%	99.5%	
MAT1A	100%	100%	100%	100%	99.2%	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850;Methionine adenosyltransferase deficiency, autosomal recessive, 250850
MAX	100%	100%	100%	100%	99.8%	Polydactyly-macrocephaly syndrome, 620712;{Pheochromocytoma, susceptibility to}, 171300
MBD5	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 1, 156200

MBOAT7	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal recessive 57, 617188
MBTPS2	94.6%	94.6%	99%	90.9%	71.9%	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	94.6%	94.5%	100%	100%	99.8%	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200
MCCC2	93.5%	93.5%	100%	100%	99.6%	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210
MCM3AP	100%	100%	100%	100%	99.7%	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development, 618124
MCM6	100%	100%	100%	100%	99.7%	Lactase persistence/nonpersistence, 223100
MCOLN1	99.1%	96.5%	100%	99.9%	99.2%	Lisch epithelial corneal dystrophy, 620763;Mucolipidosis IV, 252650
MCPH1	95%	93.6%	100%	100%	99.8%	Microcephaly 1, primary, autosomal recessive, 251200
MDGA2	100%	100%	100%	100%	99.5%	
MDH2	100%	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 51, 617339

MECP2	100%	100%	97.5%	83.5%	65.7%	Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750
MECR	93.5%	93.5%	100%	100%	99.4%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282;Optic atrophy 16, 620629
MED11	100%	100%	100%	100%	99.7%	Neurodegeneration with developmental delay, early respiratory failure, myoclonic seizures, and brain abnormalities, 620327
MED12	100%	100%	98.3%	86.8%	66.7%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED12L	100%	100%	100%	100%	99.8%	Nizon-Isidor syndrome, 618872
MED13	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 61, 618009
MED13L	100%	100%	100%	99.9%	99.3%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789

MED16	100%	100%	100%	99.6%	97.1%	Guillouet-Gordon syndrome, 621220
MED17	100%	100%	100%	100%	99.8%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668
MED23	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249
MED25	100%	100%	100%	100%	98.7%	Basel-Vanagaite-Smirin-Yosef syndrome, 616449
MED27	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286
MEF2C	100%	100%	100%	100%	99.8%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEIS2	89.6%	89.5%	100%	99.9%	99.5%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
METTL23	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 44, 615942
METTL5	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 72, 618665

MFF	89.1%	89.1%	100%	100%	99.6%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD2A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486
MFSD8	100%	100%	100%	100%	99.7%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MGAT2	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type IIa, 212066
MGP	100%	100%	100%	100%	99.2%	Keutel syndrome, 245150
MIA3	100%	100%	100%	100%	99.8%	?Ondontochondrodysplasia 2 with hearing loss and diabetes, 619269
MICOS13	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 37, 618329
MICU1	86.8%	86.7%	100%	100%	99.6%	Myopathy with extrapyramidal signs, 615673
MID1	100%	100%	99.1%	87.8%	67.2%	Opitz GBBB syndrome, 300000
MID2	100%	100%	99%	89.8%	71.7%	?Intellectual developmental disorder, X-linked 101, 300928
MIDEAS	100%	100%	100%	99.9%	99%	
MINPP1	100%	100%	100%	100%	99.8%	{Thyroid carcinoma, follicular}, 188470;Pontocerebellar hypoplasia, type 16, 619527
MIR17HG						

MKKS	100%	100%	100%	100%	99.9%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	99.5%	99%	100%	100%	99.5%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MLC1	100%	100%	100%	100%	99.6%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MLYCD	100%	100%	100%	100%	99.1%	Malonyl-CoA decarboxylase deficiency, 248360
MMAA	100%	100%	100%	100%	99.8%	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100
MMAB	92.2%	91.8%	100%	100%	99.7%	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110
MMACHC	100%	100%	100%	99.9%	99.3%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMADHC	79.3%	79.3%	100%	100%	99.7%	Methylmalonic aciduria and homocystinuria, cblD type, 277410;Methylmalonic aciduria, cblD type, 620953;Homocystinuria -megaloblastic anemia, cblD type, 620952
MMGT1	100%	100%	99.6%	90.4%	71.4%	
MMUT	100%	100%	100%	100%	99.8%	Methylmalonic aciduria, mut(0) type, 251000
MN1	100%	100%	100%	100%	99.1%	CEBALID syndrome, 618774;Meningioma, 607174
MOCS1	100%	100%	100%	100%	99.1%	Molybdenum cofactor deficiency A, 252150

MOCS2	100%	100%	100%	100%	99.9%	Molybdenum cofactor deficiency B1, 252160
MOGS	100%	100%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIb, 606056
MORC2	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688;Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090
MPDU1	100%	100%	100%	100%	99.5%	Congenital disorder of glycosylation, type If, 609180
MPDZ	100%	100%	100%	100%	99.7%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MPLKIP	100%	100%	100%	100%	99.6%	Trichothiodystrophy 4, nonphotosensitive, 234050
MPV17	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400;Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810
MRAS	100%	100%	100%	100%	99.6%	Noonan syndrome 11, 618499
MRPL49	100%	100%	100%	99.9%	99.3%	Combined oxidative phosphorylation deficiency 60, 621195
MRPS22	88.2%	86.6%	100%	100%	99.8%	Ovarian dysgenesis 7, 618117;Combined oxidative phosphorylation deficiency 5, 611719
MRPS34	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 32, 617664

MRTFB	100%	100%	100%	100%	99.6%	
MSL2	92.7%	92.7%	100%	100%	99.7%	Karayol-Borroto-Haghshenas neurodevelopmental syndrome, 620985
MSL3	100%	100%	98.8%	89.1%	70.5%	Basilicata-Akhtar syndrome, 301032
MSMO1	100%	100%	100%	100%	99.8%	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834
MSTO1	100%	100%	99.9%	99.1%	96.4%	Myopathy, mitochondrial, and ataxia, 617675
MTFMT	91.8%	91.8%	100%	100%	99%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	100%	100%	100%	100%	99.3%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTHFS	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTO1	97.8%	93.5%	100%	100%	99.9%	Combined oxidative phosphorylation deficiency 10, 614702
MTOR	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638

MTR	100%	100%	100%	100%	99.7%	{Neural tube defects, folate-sensitive, susceptibility to}, 601634;Homocystinuria -megaloblastic anemia, cblG complementation type, 250940
MTRFR	100%	100%	100%	99.9%	99.2%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTRR	100%	100%	100%	100%	99.7%	Homocystinuria-megalo blastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
MTSS2	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with ocular anomalies and distinctive facial features, 620086
MVK	100%	100%	100%	100%	99.5%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYCBP2	100%	100%	100%	100%	99.8%	
MYCN	100%	100%	100%	100%	98.9%	Feingold syndrome 1, 164280;Megalencephal y-polydactyly syndrome, 620748
MYH10	99.5%	99.5%	100%	100%	99.5%	
MYH9	97.4%	97.1%	100%	99.9%	99.4%	Macrothrombocytopeni a and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO5A	99.1%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450

MYO9A	100%	100%	100%	100%	99.9%	Myasthenic syndrome, congenital, 24, presynaptic, 618198
MYT1L	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 39, 616521
NAA10	100%	100%	98%	85.5%	68.1%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NAA15	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 50, with behavioral abnormalities, 617787
NAA20	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 73, 619717
NAA60	100%	100%	100%	100%	99.5%	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786
NACC1	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NAE1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and ischiopubic hypoplasia, 620210
NAGA	91.4%	88.1%	100%	100%	99.3%	Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241

NAGLU	100%	100%	100%	100%	99.7%	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491;Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920
NALCN	100%	100%	100%	100%	99.8%	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NANS	94.5%	94.3%	100%	100%	99.9%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NAPB	100%	100%	100%	100%	99.9%	Developmental and epileptic encephalopathy 107, 620033
NARS1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with microcephaly, impaired language, epilepsy, and gait abnormalities, autosomal dominant, 619092;Neurodevelopmental disorder with microcephaly, impaired language, and gait abnormalities, autosomal recessive, 619091
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAV3	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with poor or absent speech, dysmorphic facies, and behavioral abnormalities, 621182

NAXE	98.6%	94.5%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	97.7%	97.6%	100%	100%	99.5%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBN	97.5%	97.5%	100%	100%	99.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NCAPG2	100%	100%	100%	100%	99.7%	Khan-Khan-Katsanis syndrome, 618460
NCDN	99.6%	97.6%	100%	100%	99.3%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NCKAP1	100%	100%	100%	100%	99.9%	
NCOR2	100%	100%	100%	100%	99%	
NDC1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with achalasia, polyneuropathy, and alacrima, 621328
NDE1	93.5%	93.5%	100%	100%	98.4%	Microhydranencephaly, 605013;Lissencephaly 4 (with microcephaly), 614019
NDP	100%	100%	99.1%	90.7%	72.5%	Exudative vitreoretinopathy 2, X-linked, 305390;Norrie disease, 310600
NDST1	100%	100%	100%	99.9%	99.4%	Intellectual developmental disorder, autosomal recessive 46, 616116
NDUFA1	100%	100%	98.8%	90.4%	68.3%	Mitochondrial complex I deficiency, nuclear type 12, 301020

NDUFA11	96.1%	96%	100%	100%	98.8%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFA12	80.4%	80.4%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 23, 618244
NDUFA2	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 13, 618235
NDUFA8	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 37, 619272
NDUFAF2	67.4%	67.4%	100%	99.9%	99.6%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF5	90.9%	90.9%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFAF8	100%	100%	100%	100%	99.8%	Mitochondrial complex I deficiency, nuclear type 34, 618776
NDUFS1	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	99.1%	100%	99.9%	99.6%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	99.6%	95.6%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	98.1%	90.3%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 9, 618232

NDUFS7	89.5%	86.2%	100%	100%	99.2%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NDUFS8	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 2, 618222
NDUFV1	100%	100%	100%	99.6%	97.1%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NEDD4L	100%	100%	100%	99.9%	99.3%	Periventricular nodular heterotopia 7, 617201
NEMF	100%	100%	100%	99.6%	98.6%	Intellectual developmental disorder with speech delay and axonal peripheral neuropathy, 619099
NEU1	100%	100%	100%	100%	99.8%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEUROD2	100%	100%	100%	99.7%	96.3%	Developmental and epileptic encephalopathy 72, 618374
NEUROG1	100%	100%	100%	99.9%	97.8%	Cranial dysinnervation disorder, congenital, with absent corneal reflex and developmental delay, 620469
NEXMIF	100%	100%	98.7%	89.5%	70.8%	Intellectual developmental disorder, X-linked 98, 300912

NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFASC	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356
NFE2L2	81.2%	81.2%	100%	100%	99.5%	Immunodeficiency, developmental delay, and hypohomocysteinemia, 617744
NFIA	92.2%	92.2%	100%	100%	99%	Brain malformations with or without urinary tract defects, 613735
NFIB	100%	100%	100%	100%	99.6%	Macrocephaly, acquired, with impaired intellectual development, 618286
NFIX	100%	99.9%	100%	100%	99.1%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NFU1	99.9%	98%	100%	100%	99.9%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711
NGLY1	99.4%	97.5%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273
NHLRC2	100%	100%	100%	100%	99.8%	FINCA syndrome, 618278
NHS	100%	100%	98.9%	89.3%	69.2%	Cataract 40, X-linked, 302200;Nance-Horan syndrome, 302350

NIPBL	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 1, 122470
NKAP	100%	100%	99.1%	89.5%	69.4%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NKX2-1	100%	100%	100%	100%	99.3%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX6-2	100%	100%	100%	99.9%	98.1%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NLGN2	100%	100%	100%	99.8%	98.5%	
NLGN3	99.6%	98.3%	97.3%	85%	66.3%	{Autism susceptibility, X-linked 1}, 300425
NLGN4X	100%	100%	97.8%	86.6%	67.2%	Intellectual developmental disorder, X-linked, 300495;{Autism susceptibility, X-linked 2}, 300495
NONO	96.7%	90.7%	99.1%	90.1%	68.4%	Intellectual developmental disorder, X-linked syndromic 34, 300967
NOVA2	100%	100%	99.9%	97.9%	93.5%	Neurodevelopmental disorder with or without autistic features and/or structural brain abnormalities, 618859
NPC1	100%	100%	100%	100%	99.5%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220

NPC2	99.5%	95.6%	100%	100%	99.4%	Niemann-pick disease, type C2, 607625
NPHP1	100%	100%	100%	99.9%	99.3%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NR2F1	100%	100%	100%	99.8%	97.9%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR2F2	100%	100%	100%	99.9%	98.8%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR4A2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NRAS	89.6%	89.4%	100%	100%	99.2%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500

NRCAM	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833
NRDC	96.9%	96.9%	100%	100%	99.8%	
NRROS	100%	100%	100%	100%	99%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	100%	100%	99.7%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
NSD2	99.5%	99.5%	100%	100%	99.5%	Rauch-Steindl syndrome, 619695
NSDHL	96%	95.9%	99%	89.2%	69.5%	CK syndrome, 300831;CHILD syndrome, 308050
NSF	100%	100%	99.9%	98.6%	93.9%	Developmental and epileptic encephalopathy 96, 619340
NSRP1	91%	91%	100%	100%	99.9%	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001
NSUN2	98.5%	98.5%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 5, 611091
NSUN6	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 82, 620779
NT5C2	91.4%	91.4%	100%	100%	99.8%	Spastic paraplegia 45, autosomal recessive, 613162

NTNG2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718
NTRK1	100%	100%	100%	99.9%	98.8%	Insensitivity to pain, congenital, with anhidrosis, 256800
NTRK2	89.5%	89.5%	100%	100%	99.7%	Developmental and epileptic encephalopathy 58, 617830;Obesity, hyperphagia, and developmental delay, 613886
NUBPL	89.8%	89.8%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUDT2	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with or without peripheral neuropathy, 619844
NUP107	97.8%	97.8%	100%	100%	99.7%	?Ovarian dysgenesis 6, 618078;Galloway-Mowat syndrome 7, 618348;Nephrotic syndrome, type 11, 616730
NUP133	97.9%	97.9%	100%	100%	99.8%	?Galloway-Mowat syndrome 8, 618349;Nephrotic syndrome, type 18, 618177
NUP188	95.4%	95.4%	100%	100%	99.5%	Sandestig-Stefanova syndrome, 618804
NUP214	100%	100%	100%	100%	99.5%	Leukemia, T-cell acute lymphoblastic, somatic, 613065;Leukemia, acute myeloid, somatic, 601626;{Encephalopathy, acute, infection-induced, susceptibility to, 9}, 618426

NUP54	100%	100%	100%	100%	99.8%	Dystonia 37, early-onset, with striatal lesions, 620427
NUP62	100%	100%	100%	99.9%	98.1%	Striatonigral degeneration, infantile, 271930
NUP85	100%	100%	100%	100%	99.3%	Nephrotic syndrome, type 17, 618176
NUS1	93.9%	93.9%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
OAT	100%	100%	100%	100%	99.9%	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870
OCLN	94.5%	94.5%	100%	99.9%	98.9%	Pseudo-TORCH syndrome 1, 251290
OCRL	100%	100%	99.1%	89.6%	70.8%	Dent disease 2, 300555;Lowe syndrome, 309000
ODC1	100%	100%	100%	100%	99.8%	Bachmann-Bupp syndrome, 619075
OFD1	100%	100%	99.2%	90.8%	71.1%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDH	100%	100%	100%	100%	99.4%	Oxoglutarate dehydrogenase deficiency, 203740
OGDHL	100%	100%	100%	100%	99.5%	
OGT	100%	100%	99.4%	91.5%	73%	Intellectual developmental disorder, X-linked 106, 300997

OLA1	100%	100%	100%	100%	99.7%	
OPA3	100%	100%	100%	100%	99.7%	3-methylglutaconic aciduria, type III, 258501;Optic atrophy 3 with cataract, 165300
OPHN1	94%	94%	99.1%	89.5%	69.7%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
ORC1	100%	100%	100%	100%	99.4%	Meier-Gorlin syndrome 1, 224690
OSGEP	100%	100%	100%	100%	99.6%	Galloway-Mowat syndrome 3, 617729
OTC	100%	100%	99.1%	90.6%	73%	Ornithine transcarbamylase deficiency, 311250
OTUD5	100%	100%	98.9%	87.8%	67.1%	Multiple congenital anomalies-neurodevelopmental syndrome, X-linked, 301056
OTUD6B	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452
OTUD7A	100%	98.7%	100%	99.9%	98%	Neurodevelopmental disorder with hypotonia and seizures, 620790
OTX2	100%	100%	100%	100%	99.8%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
OXR1	100%	100%	100%	100%	99.9%	Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay, 213000

P4HTM	89.4%	89.4%	100%	100%	99.2%	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities, 618493
PABPC1	94.4%	93.8%	100%	100%	99.9%	
PACS1	100%	100%	100%	100%	99.3%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	78.8%	78.8%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100%	100%	100%	100%	99.8%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432
PAH	96.7%	94.1%	100%	100%	99.8%	[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PAK1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder with macrocephaly, seizures, and speech delay, 618158
PAK3	100%	100%	99.1%	90.2%	71.7%	Intellectual developmental disorder, X-linked 30, 300558
PALS1	100%	100%	100%	100%	99.9%	
PAM16	86.6%	85.5%	100%	100%	99.5%	Spondylometaphyseal dysplasia, Megarbane-Dagher-Me like type, 613320
PAN2	100%	100%	100%	100%	99.6%	
PANK2	100%	100%	100%	100%	99.8%	Neurodegeneration with brain iron accumulation 1, 234200

PANX1	100%	100%	100%	100%	99.7%	Oocyte/zygote/embryo maturation arrest 7, 618550
PARN	98.3%	96.3%	100%	100%	99.9%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP6	100%	100%	100%	99.9%	99.5%	
PARS2	100%	100%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 75, 618437
PAX1	100%	100%	100%	99.9%	98.2%	Otofaciocervical syndrome 2 with T-cell deficiency, 615560
PAX7	94.2%	94.2%	100%	100%	99.3%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PAX8	100%	100%	100%	100%	99.3%	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700
PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PC	100%	99.3%	100%	100%	99.2%	Pyruvate carboxylase deficiency, 266150
PCCA	84.6%	84.4%	100%	100%	99.7%	Propionicacidemia, 606054
PCCB	86.8%	85.3%	100%	100%	99.7%	Propionicacidemia, 606054

PCDH12	100%	100%	100%	100%	99.3%	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280
PCDH19	100%	100%	97.3%	84.8%	64.4%	Developmental and epileptic encephalopathy 9, 300088
PCDHGC4	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with poor growth and skeletal anomalies, 619880
PCGF2	100%	100%	100%	100%	98.6%	Turnpenny-Fry syndrome, 618371
PCLO	100%	100%	100%	99.9%	99.3%	Pontocerebellar hypoplasia, type 3, 608027
PCNT	100%	100%	100%	100%	99.6%	Microcephalic osteodysplastic primordial dwarfism, type II, 210720
PCYT2	100%	99%	100%	100%	99.6%	Spastic paraplegia 82, autosomal recessive, 618770
PDCD6IP	100%	100%	100%	100%	99.8%	?Microcephaly 29, primary, autosomal recessive, 620047
PDE2A	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE4D	100%	100%	100%	100%	99.6%	Acrodysostosis 2, with or without hormone resistance, 614613

PDGFRB	99.8%	98.1%	100%	100%	99.3%	Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PDHA1	91.7%	89.7%	98.9%	88.8%	69.2%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100%	100%	100%	100%	99.6%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	91.6%	91.6%	100%	100%	99.7%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100%	100%	100%	100%	99.6%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDSS1	100%	100%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PDZD8	100%	100%	100%	99.7%	98.4%	Intellectual developmental disorder with autism and dysmorphic facies, 620021
PEPD	94%	94%	100%	100%	99.6%	Prolidase deficiency, 170100
PET100	100%	100%	100%	100%	99.5%	Mitochondrial complex IV deficiency, nuclear type 12, 619055

PEX1	100%	100%	100%	100%	99.7%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100%	100%	100%	100%	99.1%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX11B	100%	100%	100%	100%	99.8%	Peroxisome biogenesis disorder 14B, 614920
PEX12	100%	100%	100%	100%	99.5%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	92.4%	92.4%	100%	100%	99.8%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX16	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	88.6%	88.6%	100%	100%	99.7%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX2	100%	100%	100%	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX26	92.6%	91.7%	100%	100%	99%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

PEX3	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	99.9%	99.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PEX7	97.9%	97.9%	100%	100%	99.7%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PGAP1	96.7%	96.7%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802
PGAP2	100%	100%	100%	100%	99.6%	Hyperphosphatasia with impaired intellectual development syndrome 3, 614207
PGAP3	100%	100%	100%	100%	99.1%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PGK1	92.7%	92.7%	98.9%	89.9%	70.4%	Phosphoglycerate kinase 1 deficiency, 300653

PGM2L1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skin abnormalities, 620191
PGM3	100%	100%	100%	100%	99.8%	Immunodeficiency 23, 615816
PHACTR1	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 70, 618298
PHF21A	100%	100%	100%	100%	99.6%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF5A	85%	85%	100%	100%	99.8%	
PHF6	100%	100%	99.4%	91.3%	72.1%	Borjeson-Forssman-Lehmann syndrome, 301900
PHF8	100%	100%	98.7%	88.9%	69.1%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263
PHGDH	100%	100%	100%	99.9%	99.3%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHIP	99.4%	99.4%	100%	100%	99.6%	Chung-Jansen syndrome, 617991
PI4K2A	86.9%	86.9%	100%	100%	99.2%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732

PI4KA	99.3%	99.2%	100%	99.8%	98.8%	Spastic paraplegia 84, autosomal recessive, 619621;Gastrointestinal defects and immunodeficiency syndrome 2, 619708;Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531
PIBF1	100%	100%	100%	100%	99.6%	Joubert syndrome 33, 617767
PIDD1	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 75, with neuropsychiatric features and variant lissencephaly, 619827
PIGA	100%	100%	99.5%	90.7%	71.6%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGB	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 80, 618580
PIGC	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 16, 617816
PIGF	100%	100%	100%	100%	99.9%	Onychodystrophy, osteodystrophy, impaired intellectual development, and seizures syndrome, 619356

PIGG	100%	100%	100%	100%	99.6%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGH	90.2%	80.2%	100%	100%	99.1%	Glycosylphosphatidylinositol biosynthesis defect 17, 618010
PIGK	88.2%	88.2%	100%	100%	99.9%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGL	100%	100%	100%	100%	99.8%	CHIME syndrome, 280000
PIGN	97%	95%	100%	100%	99.9%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	100%	99.5%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	100%	99.6%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548
PIGS	99.5%	97%	100%	100%	99.3%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	100%	100%	99.5%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398

PIGU	97.1%	97.1%	100%	100%	99.9%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PIGV	100%	100%	100%	100%	99.6%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PIGW	100%	100%	100%	100%	99.8%	Glycosylphosphatidylinositol biosynthesis defect 11, 616025
PIGY	100%	100%	100%	100%	99.1%	Hyperphosphatasia with impaired intellectual development syndrome 6, 616809
PIK3C2A	100%	100%	100%	100%	99.8%	Oculoskeletodental syndrome, 618440

PIK3CA	100%	100%	100%	100%	99.6%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3R2	100%	100%	100%	99.9%	99.1%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387
PIP5K1C	91.3%	90.3%	100%	100%	99.5%	Lethal congenital contractural syndrome 3, 611369
PISD	100%	100%	100%	100%	99.8%	Liberfarb syndrome, 618889
PITRM1	100%	100%	100%	100%	99.5%	Spinocerebellar ataxia, autosomal recessive 30, 619405
PJA1	100%	100%	97.8%	83.3%	63.9%	

PLA2G6	100%	100%	100%	100%	99.6%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLAA	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527
PLAAT3	67.6%	63.2%	100%	100%	99.7%	Lipodystrophy, familial partial, type 9, 620683
PLCB1	100%	100%	100%	100%	99.8%	Developmental and epileptic encephalopathy 12, 613722
PLEKHG2	100%	100%	100%	100%	99.3%	Leukodystrophy and acquired microcephaly with or without dystonia, 616763
PLK1	100%	100%	100%	99.9%	99%	
PLK4	100%	100%	100%	100%	99.8%	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171
PLP1	96.1%	94.3%	99%	89.8%	69.2%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PLPBP	100%	100%	100%	100%	99.1%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PLXNA1	100%	100%	100%	100%	99.4%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PLXNA2	100%	100%	100%	100%	99.5%	
PLXNB2	100%	100%	100%	99.9%	98.7%	

PLXND1	100%	100%	100%	100%	99.3%	Congenital heart defects, multiple types, 9, 620294
PMM2	94.6%	94.6%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	96.2%	96.2%	100%	100%	99.3%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PMPCB	91.4%	91.4%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 6, 617954
PNKP	100%	100%	100%	99.9%	98.7%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNP	100%	100%	100%	100%	99.8%	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179
PNPLA6	100%	100%	100%	99.9%	99.1%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
PNPLA8	100%	100%	100%	100%	99.8%	Mitochondrial myopathy with lactic acidosis, 251950
PNPO	100%	100%	100%	100%	99.3%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090

PNPT1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POGZ	100%	100%	100%	100%	99%	White-Sutton syndrome, 616364
POLA1	100%	100%	99.2%	91.1%	71.9%	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030
POLG	100%	100%	100%	100%	99.4%	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
POLR1A	100%	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	84.4%	83.5%	100%	99.9%	99%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390

POLR2A	100%	100%	100%	99.8%	98.7%	Neurodevelopmental disorder with hypotonia and variable intellectual and behavioral abnormalities, 618603
POLR3A	100%	100%	100%	100%	99.4%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3K	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 21, 619310
POLRMT	99.9%	99.2%	100%	100%	99.2%	Combined oxidative phosphorylation deficiency 55, 619743
POMGNT1	100%	100%	100%	100%	99.7%	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%	100%	100%	100%	99.5%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830;Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135
POMK	100%	100%	100%	100%	99.6%	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249
POMT1	100%	99.8%	100%	100%	99.4%	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	95.1%	94.9%	100%	99.8%	98.7%	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%	100%	98.5%	86.2%	67.6%	Focal dermal hypoplasia, 305600

POU1F1	100%	100%	100%	100%	99.6%	Pituitary hormone deficiency, combined or isolated, 1, 613038
POU3F2	100%	100%	100%	99.9%	98.1%	
POU3F3	100%	99.5%	99.8%	98.1%	89.8%	Snijders Blok-Fisher syndrome, 618604
POU4F1	95.6%	92.5%	100%	99.7%	96.2%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
PPFIA2	100%	100%	100%	100%	99.8%	
PPFIA3	100%	100%	100%	100%	99%	Paul-Chao neurodevelopmental syndrome, 621122
PPFIBP1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPIL1	80.1%	80.1%	100%	100%	99.2%	Pontocerebellar hypoplasia, type 14, 619301
PPM1D	100%	100%	100%	99.9%	99.6%	Breast cancer, somatic, 114480;Jansen-de Vries syndrome, 617450
PPP1CB	88.1%	87.6%	100%	100%	99.8%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PPP1R12A	100%	100%	100%	100%	99.9%	Genitourinary and/or/brain malformation syndrome, 618820
PPP1R15B	100%	100%	100%	100%	99.9%	Microcephaly, short stature, and impaired glucose metabolism 2, 616817
PPP1R21	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities, 619383

PPP1R3F	97%	97%	98.5%	90.1%	70.5%	
PPP2CA	100%	100%	100%	100%	99.8%	Houge-Janssens syndrome 3, 618354
PPP2R1A	94%	94%	100%	100%	99.8%	Houge-Janssens syndrome 2, 616362
PPP2R2B	100%	100%	100%	100%	99.3%	Spinocerebellar ataxia 12, 604326
PPP2R3C	100%	100%	100%	100%	100%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PPP2R5B	100%	100%	100%	100%	99.5%	
PPP2R5C	99.4%	97.6%	100%	100%	99.3%	Houge-Janssens syndrome 4, 621185
PPP2R5D	100%	100%	100%	100%	99.2%	Houge-Janssens syndrome 1, 616355
PPP3CA	100%	100%	100%	100%	99.6%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PPT1	90.3%	90.3%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	98.5%	86.1%	66.5%	Renpenning syndrome, 309500
PRDM13	100%	100%	100%	100%	98.9%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761
PRDM15	100%	100%	100%	100%	99.5%	
PRICKLE2	100%	100%	100%	100%	99%	
PRKACB	94.2%	94.2%	100%	100%	99.7%	Cardioacrofacial dysplasia 2, 619143

PRKAR1A	100%	100%	100%	100%	99.6%	Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960
PRKAR1B	100%	100%	100%	100%	98.7%	Marbach-Schaaf neurodevelopmental syndrome, 619680
PRMT7	100%	100%	100%	100%	99.4%	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157
PRMT9	97.7%	97.7%	99.9%	99%	96.5%	
PRODH	92.2%	91.9%	100%	100%	99.5%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500
PRPF19	100%	100%	100%	100%	99.4%	
PRPF8	100%	100%	100%	100%	99.2%	Retinitis pigmentosa 13, 600059
PRPS1	100%	100%	99%	88.4%	69.2%	Arts syndrome, 301835;Phosphoribosyl pyrophosphate synthetase superactivity, 300661;Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070;Deafness, X-linked 1, 304500;Gout, PRPS-related, 300661
PRR12	100%	100%	100%	99.9%	98.4%	Neuroocular syndrome, 619539
PRSS12	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal recessive 1, 249500

PRUNE1	93.1%	93.1%	100%	99.9%	99.2%	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481
PSAP	100%	98.6%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PSAT1	100%	100%	100%	100%	99.7%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSMC3	100%	100%	100%	100%	99%	?Deafness, cataract, impaired intellectual development, and polyneuropathy, 619354
PSMC5	100%	100%	100%	100%	99.6%	
PSMD11	94.1%	94.1%	100%	100%	99.8%	
PSMD12	89.3%	89.3%	100%	100%	99.5%	Stankiewicz-Isidor syndrome, 617516
PSPH	100%	100%	100%	100%	99.6%	Phosphoserine phosphatase deficiency, 614023
PTBP1	100%	99.9%	100%	99.9%	99.2%	
PTCD3	100%	100%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 51, 619057

PTCH1	100%	100%	100%	100%	99.1%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTCHD1	100%	100%	99%	90%	70.4%	{Autism, susceptibility to, X-linked 4}, 300830
PTDSS1	93%	93%	100%	100%	99.8%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	89.6%	89.6%	100%	100%	99.2%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTF1A	100%	100%	100%	99.9%	98.7%	Pancreatic and cerebellar agenesis, 609069;Pancreatic agenesis 2, 615935
PTPA	100%	100%	100%	100%	99.4%	Parkinson disease 25, autosomal recessive early-onset, with impaired intellectual development, 620482
PTPMT1	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with ataxia and brain abnormalities, 621199
PTPN11	90.5%	89.2%	100%	100%	99.6%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785

PTPN23	100%	100%	100%	99.9%	98.8%	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890
PTPN4	100%	100%	100%	100%	99.9%	
PTRH2	100%	100%	100%	100%	100%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTRHD1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with early-onset parkinsonism and behavioral abnormalities, 620747
PTS	100%	100%	100%	100%	99.7%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUF60	100%	100%	100%	100%	99.5%	Verheij syndrome, 615583
PUM1	92.4%	92.4%	100%	99.8%	98.9%	Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719
PURA	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PUS1	100%	100%	100%	99.9%	99.4%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
PUS3	100%	100%	100%	100%	100%	Neurodevelopmental disorder with microcephaly and gray sclerae, 617051

PUS7	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with abnormal behavior, microcephaly, and short stature, 618342
PYCR1	100%	100%	100%	100%	99.1%	Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940
PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100%	100%	100%	100%	99.5%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	93.7%	93.6%	100%	100%	99.5%	Hyperphenylalaninemia, BH4-deficient, C, 261630
QRICH1	100%	100%	100%	100%	99.6%	Ververi-Brady syndrome, 617982
RAB11A	92.9%	92.9%	100%	100%	99.8%	
RAB11B	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807
RAB14	100%	100%	100%	100%	99.6%	
RAB18	100%	100%	100%	100%	99.5%	Warburg micro syndrome 3, 614222
RAB23	100%	100%	100%	100%	99.8%	Carpenter syndrome, 201000
RAB27A	100%	100%	100%	99.9%	99.3%	Griscelli syndrome, type 2, 607624
RAB34	100%	100%	100%	100%	99.5%	Orofaciodigital syndrome XX, 620718

RAB39B	100%	100%	99.3%	90.4%	69.8%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RAB3GAP1	95.7%	95.7%	100%	100%	99.6%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	94.5%	94.4%	100%	100%	99.8%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RAB5C	100%	100%	100%	99.6%	98.4%	
RABGAP1	91.5%	90.5%	100%	100%	99.7%	
RAC1	78.8%	78.8%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAC3	98.8%	92.8%	100%	100%	99.3%	Neurodevelopmental disorder with structural brain anomalies and dysmorphic facies, 618577
RAD21	100%	100%	100%	100%	99.7%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAF1	98.5%	95.2%	100%	100%	99.8%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RAI1	100%	100%	100%	100%	99.2%	Smith-Magenis syndrome, 182290
RALA	100%	100%	100%	100%	99.7%	Hiatt-Neu-Cooper neurodevelopmental syndrome, 619311
RALGAPA1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation, 618797

RAP1B	100%	100%	100%	100%	99.7%	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654
RAPGEF2	100%	100%	100%	100%	99.9%	?Epilepsy, familial adult myoclonic, 7, 618075
RARB	98%	98%	100%	100%	99.9%	Microphthalmia, syndromic 12, 615524
RARS1	93.4%	90.7%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	94.2%	94.2%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 6, 611523
RBBP5	100%	100%	100%	100%	99.6%	
RBBP8	100%	100%	100%	100%	99.9%	Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBFOX1	100%	99.8%	100%	99.9%	99.4%	
RBL2	100%	100%	100%	100%	99.8%	Brunet-Wagner neurodevelopmental syndrome, 619690
RBM10	95.6%	95.6%	97.9%	85.5%	65.9%	TARP syndrome, 311900
RBM28	100%	100%	100%	100%	99.6%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBPJ	100%	100%	100%	100%	99.9%	Adams-Oliver syndrome 3, 614814
RBSN	96.1%	96.1%	100%	100%	99.7%	Myelofibrosis, congenital, with anemia, neutropenia, developmental delay, and ocular abnormalities, 620939;Kariminejad neurodevelopmental syndrome, 620937

RCBTB1	100%	100%	100%	100%	99.9%	Retinal dystrophy with or without extraocular anomalies, 617175
RECQL4	100%	100%	100%	100%	99.1%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RELN	100%	100%	100%	100%	99.7%	{Epilepsy, familial temporal lobe, 7}, 616436;Lissencephaly 2 (Norman-Roberts type), 257320
RERE	99%	99%	100%	99.7%	98.1%	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975
REV3L	100%	100%	100%	100%	99.8%	
RFC4	100%	100%	100%	100%	99.6%	Morimoto-Ryu-Malicdan neuromuscular syndrome, 621010
RFT1	84.9%	84.9%	100%	100%	99.8%	Congenital disorder of glycosylation, type In, 612015
RFX3	100%	100%	100%	100%	99.8%	
RFX4	100%	100%	100%	99.7%	98.8%	
RFX7	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 71, with behavioral abnormalities, 620330
RHEB	100%	100%	100%	100%	99.6%	
RHOBTB2	98.7%	98.7%	100%	99.9%	99.5%	Developmental and epileptic encephalopathy 64, 618004
RIC1	100%	100%	100%	100%	99.9%	CATIFA syndrome, 618761
RICTOR	100%	100%	100%	100%	99.9%	

RIMS2	99.2%	99.2%	100%	100%	99.7%	Cone-rod synaptic disorder syndrome, congenital nonprogressive, 618970
RIT1	100%	100%	100%	100%	99.9%	Noonan syndrome 8, 615355
RLF	100%	100%	100%	100%	99.8%	
RLIM	100%	100%	99.2%	88.3%	67.6%	Tonne-Kalscheuer syndrome, 300978
RMND1	85.8%	85.8%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 11, 614922
RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNASEH2A	100%	100%	100%	100%	99.4%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.5%	91.5%	100%	100%	99.6%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	100%	99.4%	Aicardi-Goutieres syndrome 3, 610329
RNASET2	82%	77.9%	100%	100%	99.7%	Leukoencephalopathy, cystic, without megalencephaly, 612951
RNF113A	100%	100%	97.6%	86.6%	67.6%	Trichothiodystrophy 5, nonphotosensitive, 300953
RNF125	100%	100%	100%	100%	99.5%	Tenorio syndrome, 616260
RNF13	85.5%	85.3%	100%	100%	99.7%	Developmental and epileptic encephalopathy 73, 618379
RNF2	100%	100%	100%	100%	99.4%	Luo-Schoch-Yamamoto syndrome, 619460

RNF220	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688
RNPC3	100%	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 7, 618160
RNU12						CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208
RNU2-2						Developmental and epileptic encephalopathy 119, 621304
RNU4-2						ReNU syndrome, 620851
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RNU5B-1						Neurodevelopmental disorder with seizures and joint laxity, 621302
RNU7-1						Aicardi-Goutieres syndrome 9, 619487
ROBO1	100%	100%	100%	100%	99.8%	Pituitary hormone deficiency, combined or isolated, 8, 620303;Neurooculorenal syndrome, 620305;?Nystagmus 8, congenital, autosomal recessive, 257400
ROGDI	100%	100%	100%	100%	99.3%	Kohlschutter-Tonz syndrome, 226750
ROR2	100%	100%	100%	100%	99.6%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310

RORA	100%	100%	100%	100%	99.1%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RORB	100%	100%	100%	100%	99.9%	{Epilepsy, idiopathic generalized, susceptibility to, 15}, 618357
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPH3A	100%	100%	100%	99.9%	99.2%	
RPIA	100%	100%	100%	100%	99.8%	Ribose 5-phosphate isomerase deficiency, 608611
RPL10	100%	100%	98.6%	89.2%	69.8%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RPS19	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 1, 105650
RPS6KA3	97.6%	97.6%	99.2%	92.2%	73%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600
RPS6KC1	97.6%	97.6%	100%	100%	99.7%	
RRAS2	95.5%	95.5%	100%	100%	99.8%	Ovarian carcinoma;Noonan syndrome 12, 618624

RRM2B	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
RRP7A	100%	100%	100%	99.6%	97.7%	?Microcephaly 28, primary, autosomal recessive, 619453
RSF1	100%	100%	100%	99.9%	99.6%	
RSPRY1	95.5%	95.5%	100%	100%	99.5%	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type, 616723
RSRC1	84.2%	84.2%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 70, 618402
RTEL1	100%	100%	100%	100%	99.5%	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%	100%	100%	100%	99.9%	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732
RTTN	100%	100%	100%	100%	99.8%	Microcephaly, short stature, and polymicrogyria with seizures, 614833
RUBCN	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 15, 615705
RUNX1T1	100%	100%	100%	99.9%	99.4%	
RUSC2	100%	100%	100%	100%	99.5%	Intellectual developmental disorder, autosomal recessive 61, 617773
RXYLT1	100%	100%	100%	100%	99.9%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041
RYBP	100%	100%	100%	100%	99.2%	
SALL1	100%	100%	100%	100%	99.3%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
SAMD9	100%	100%	100%	100%	99.9%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMHD1	94.1%	91.3%	100%	100%	99.7%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SARS1	89.1%	89.1%	100%	99.9%	99.4%	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709

SART3	100%	99.9%	100%	100%	99.6%	
SATB1	100%	100%	100%	100%	99.9%	den Hoed-de Boer-Voisin syndrome, 619229;Developmental delay with dysmorphic facies and dental anomalies, 619228
SATB2	100%	100%	100%	100%	99.7%	Glass syndrome, 612313
SBDS	100%	100%	100%	99.9%	99.6%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SC5D	100%	100%	100%	100%	100%	Lathosterolosis, 607330
SCAF4	100%	100%	100%	100%	99.4%	Fliedner-Zweier syndrome, 620511
SCAMP5	100%	100%	100%	100%	99.1%	
SCAPER	97.9%	97.9%	100%	100%	99.8%	Intellectual developmental disorder and retinitis pigmentosa, 618195
SCN1A	100%	100%	100%	100%	99.8%	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%	100%	100%	99.9%	99.3%	Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838
SCN2A	100%	100%	100%	100%	99.8%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
SCN3A	100%	100%	100%	100%	99.8%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN8A	100%	100%	100%	100%	99.6%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558
SCO1	100%	100%	100%	100%	99.7%	Mitochondrial complex IV deficiency, nuclear type 4, 619048
SCO2	100%	100%	100%	100%	99.1%	Myopia 6, 608908;Mitochondrial complex IV deficiency, nuclear type 2, 604377
SCUBE3	97.5%	97.5%	100%	100%	99.6%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 619184

SCYL1	100%	100%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SCYL2	97.3%	97.3%	100%	100%	99.9%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SDCCAG8	100%	100%	100%	100%	99.7%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SDHA	94.4%	90.5%	100%	99.9%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165
SDHAF1	100%	100%	100%	100%	99.9%	Mitochondrial complex II deficiency, nuclear type 2, 619166
SEC31A	96.7%	96.6%	100%	100%	99.6%	?Halperin-Birk syndrome, 618651
SECISBP2	100%	100%	100%	100%	99.8%	Thyroid hormone metabolism, abnormal, 1, 609698
SEMA3E	100%	100%	100%	100%	99.8%	
SEMA6B	100%	100%	100%	99.9%	99%	Epilepsy, progressive myoclonic, 11, 618876
SEPHS1	92.7%	88.5%	100%	100%	99.5%	Ververi-Brady syndrome 2, 621325
SEPSECS	99.9%	98%	100%	100%	99.5%	Pontocerebellar hypoplasia type 2D, 613811
SEPTIN2	93.9%	93.9%	100%	100%	99.9%	

SERAC1	100%	100%	100%	100%	99.8%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SET	100%	100%	100%	99.9%	99%	Intellectual developmental disorder, autosomal dominant 58, 618106
SETBP1	100%	100%	100%	100%	99.4%	Schinzel-Giedion midface retraction syndrome, 269150;Intellectual developmental disorder, autosomal dominant 29, 616078
SETD1A	100%	100%	100%	99.9%	98.8%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100%	100%	100%	99.9%	98.3%	Intellectual developmental disorder with seizures and language delay, 619000
SETD2	100%	100%	100%	100%	99.7%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SETD5	99.9%	99.3%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 23, 615761
SF1	100%	100%	100%	99.9%	98.8%	
SF3B1	100%	100%	100%	100%	99.9%	Myelodysplastic syndrome, somatic, 614286
SF3B3	98.2%	97.5%	100%	100%	99.8%	

SFXN4	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 18, 615578
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SGSH	100%	100%	100%	99.9%	99.3%	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900
SGSM3	100%	100%	100%	100%	99.4%	
SHANK1	99.2%	98.6%	100%	99.7%	97.5%	
SHANK2	100%	100%	100%	99.9%	99.2%	{Autism susceptibility 17}, 613436
SHANK3	99.8%	98.7%	99.9%	99.6%	97.7%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SHH	100%	100%	100%	100%	98.3%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SHMT2	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities, 619121
SHOC2	100%	100%	100%	100%	99.3%	Noonan syndrome-like with loose anagen hair 1, 607721
SHQ1	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SHROOM4	100%	100%	98.9%	89.7%	70%	
SIAH1	100%	100%	100%	100%	99.8%	Buratti-Harel syndrome, 619314
SIK1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 30, 616341

SIL1	91.5%	91.5%	100%	100%	99.2%	Marinesco-Sjogren syndrome, 248800
SIN3A	97.5%	97.5%	100%	100%	99.5%	Witteveen-Kolk syndrome, 613406
SIN3B	99.6%	98.1%	100%	99.9%	99.3%	
SIX3	100%	100%	100%	99.9%	98.7%	Schizencephaly, 269160;Holoprosencephaly 2, 157170
SKI	100%	100%	100%	99.9%	98.9%	Shprintzen-Goldberg syndrome, 182212
SKIC3	98.9%	98.9%	100%	100%	99.7%	Trichohepatoenteric syndrome 1, 222470
SKOR2	100%	100%	100%	99.8%	97.2%	
SLC12A2	100%	100%	100%	99.9%	99.6%	Kilquist syndrome, 619080;Delpire-McNeill syndrome, 619083;Deafness, autosomal dominant 78, 619081
SLC12A5	100%	100%	100%	100%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC12A6	100%	100%	100%	100%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC12A9	100%	100%	100%	99.9%	98.9%	
SLC13A5	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905
SLC16A2	88.4%	88.4%	98.1%	88.4%	68.7%	Allan-Herndon-Dudley syndrome, 300523

SLC17A5	92.7%	92.7%	100%	99.9%	99.4%	Salla disease, 604369;Sialic acid storage disorder, infantile, 269920
SLC18A2	99.9%	97.9%	100%	100%	99.5%	Parkinsonism-dystonia, infantile, 2, 618049
SLC19A3	100%	99.4%	100%	100%	99.7%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A1	100%	100%	100%	100%	99.9%	Dicarboxylic aminoaciduria, 222730;{?Schizophrenia susceptibility 18}, 615232
SLC1A2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 41, 617105
SLC1A4	100%	100%	100%	99.8%	99.5%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC25A1	100%	100%	100%	100%	99.1%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A12	92.7%	90.8%	100%	100%	99.8%	Developmental and epileptic encephalopathy 39, 612949
SLC25A15	100%	100%	100%	100%	99.9%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 3, 609304
SLC25A24	99.5%	99.5%	99.4%	99.1%	98.1%	Fontaine progeroid syndrome, 612289

SLC25A42	100%	100%	100%	99.9%	99.2%	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression, 618416
SLC27A3	100%	100%	100%	100%	99.2%	
SLC2A1	100%	100%	100%	100%	99.5%	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%	100%	100%	100%	99.9%	Birk-Landau-Perez syndrome, 617595
SLC31A1	100%	100%	100%	100%	99.8%	Neurodegeneration and seizures due to copper transport defect, 620306
SLC32A1	100%	100%	100%	100%	99.6%	Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482
SLC35A1	94.9%	94.9%	100%	100%	99.9%	Congenital disorder of glycosylation, type IIf, 603585
SLC35A2	100%	100%	97.9%	87.7%	66.9%	Congenital disorder of glycosylation, type IIm, 300896

SLC35A3	92%	89%	100%	100%	99.8%	Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC35B2	100%	100%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 26, with chondrodysplasia, 620269
SLC35C1	100%	100%	100%	99.8%	98.9%	Congenital disorder of glycosylation, type IIc, 266265
SLC38A3	96.3%	96.3%	100%	100%	99.6%	Developmental and epileptic encephalopathy 102, 619881
SLC39A14	94%	94%	100%	99.9%	99.6%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC39A8	77.5%	77.5%	100%	100%	99.9%	Congenital disorder of glycosylation, type IIn, 616721
SLC45A1	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with neuropsychiatric features, 617532
SLC46A1	100%	100%	100%	99.9%	98.9%	Folate malabsorption, hereditary, 229050
SLC4A10	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with hypotonia and characteristic brain abnormalities, 620746
SLC4A4	95%	95%	100%	100%	99.7%	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278
SLC5A6	100%	100%	100%	100%	99.8%	Sodium-dependent multivitamin transporter deficiency, 618973;Peripheral motor neuropathy, childhood-onset, biotin-responsive, 619903

SLC5A7	100%	100%	100%	100%	99.5%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A1	100%	100%	100%	100%	99.5%	Myoclonic-atonic epilepsy, 616421
SLC6A17	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 48, 616269
SLC6A19	100%	100%	100%	99.9%	99%	Hartnup disorder, 234500
SLC6A3	94.6%	92.6%	100%	100%	99.4%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SLC6A8	100%	100%	98.2%	86.6%	67.6%	Cerebral creatine deficiency syndrome 1, 300352
SLC6A9	100%	100%	100%	100%	99.5%	Glycine encephalopathy with normal serum glycine, 617301
SLC7A7	100%	100%	100%	99.8%	98.3%	Lysinuric protein intolerance, 222700
SLC9A6	100%	100%	99.2%	90.4%	69.9%	Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairement, 301142;Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SLC9A7	100%	100%	99%	89.6%	69%	Intellectual developmental disorder, X-linked 108, 301024

SLF2	100%	100%	100%	100%	99.7%	Atelis syndrome 1, 620184
SLITRK2	100%	100%	98.6%	87.6%	68.1%	Intellectual developmental disorder, X-linked 111, 301107
SMAD4	95.4%	95.4%	100%	100%	99.9%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA1	98%	98%	99.2%	91.1%	71.5%	
SMARCA2	98%	97.8%	100%	100%	99.7%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100%	100%	100%	100%	99.1%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCA5	98.4%	98.4%	100%	100%	99.8%	
SMARCB1	95.2%	95.2%	100%	99.9%	99.3%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCC2	94.2%	94.2%	100%	100%	99.3%	Coffin-Siris syndrome 8, 618362
SMARCD1	100%	100%	100%	100%	99.3%	Coffin-Siris syndrome 11, 618779

SMARCE1	100%	100%	100%	100%	99.9%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SMC1A	100%	100%	98.6%	87.2%	66.6%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	100%	100%	99.7%	Cornelia de Lange syndrome 3, 610759
SMC5	97.3%	97.3%	100%	100%	99.8%	Atelis syndrome 2, 620185
SMG8	100%	100%	100%	100%	99.8%	Alzahrani-Kuwahara syndrome, 619268
SMG9	100%	100%	100%	100%	99.4%	Heart and brain malformation syndrome, 616920;Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies, 619995
SMOC1	96%	96%	100%	100%	99.8%	Microphthalmia with limb anomalies, 206920
SMPD1	100%	100%	100%	100%	99.2%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SMPD4	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	96.6%	94.9%	99%	89.9%	71.2%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583

SNAP25	89.5%	89.5%	100%	100%	99.9%	Developmental and epileptic encephalopathy 117, 616330
SNAP29	100%	100%	100%	100%	99.8%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNAPC4	100%	100%	100%	100%	98.9%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515
SNAPIN	100%	100%	100%	99.7%	98.6%	
SNF8	94.6%	94.6%	100%	100%	99.6%	Developmental and epileptic encephalopathy 115, 620783;Neurodevelopmental disorder plus optic atrophy, 620784
SNIP1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501
SNORD118						Leukoencephalopathy, brain calcifications, and cysts, 614561
SNRPB	94.4%	86.2%	100%	100%	99.7%	Cerebrocostomandibular syndrome, 117650
SNRPN	100%	100%	100%	100%	99.7%	
SNX14	95%	95%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SNX27	100%	100%	100%	100%	99.7%	
SOBP	100%	100%	100%	99.5%	96.4%	?Impaired intellectual development, anterior maxillary protrusion, and strabismus, 613671

SON	100%	100%	100%	100%	99.7%	ZTTK syndrome, 617140
SOS1	98.8%	98.8%	100%	100%	99.8%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	95%	95%	100%	100%	99.3%	Noonan syndrome 9, 616559
SOX10	97.8%	97.8%	100%	99.8%	99%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100%	100%	100%	99.5%	94.3%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX2	100%	100%	100%	99.8%	97.6%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	100%	100%	96.5%	80.8%	58.8%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX4	100%	100%	100%	99.9%	98.3%	Intellectual developmental disorder with speech delay and dysmorphic facies, 618506
SOX5	100%	100%	100%	100%	99.8%	Lamb-Shaffer syndrome, 616803
SOX6	100%	100%	100%	100%	99.7%	Tolchin-Le Caignec syndrome, 618971

SP9	100%	100%	100%	99.9%	98.6%	
SPAG9	100%	100%	100%	100%	99.5%	
SPART	95.8%	95.8%	100%	100%	99.7%	Troyer syndrome, 275900
SPAST	94.7%	94.6%	100%	99.8%	98.5%	Spastic paraplegia 4, autosomal dominant, 182601
SPECC1L	94.6%	94.6%	100%	100%	99.8%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
SPEN	100%	100%	100%	100%	99.4%	Radio-Tartaglia syndrome, 619312
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPOCK1	100%	100%	100%	99.9%	99.2%	
SPOP	96.4%	96.4%	100%	100%	99.9%	Nabais Sa-de Vries syndrome, type 1, 618828;Nabais Sa-de Vries syndrome, type 2, 618829
SPOUT1	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder with poor growth, seizures, and brain abnormalities, 621154
SPR	100%	100%	100%	100%	99.4%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPRED1	97.6%	97.6%	100%	100%	99.9%	Legius syndrome, 611431
SPRED2	100%	100%	100%	100%	99.4%	Noonan syndrome 14, 619745

SPTAN1	99.5%	98.8%	100%	100%	99.5%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528
SPTBN1	100%	100%	100%	100%	99.6%	Developmental delay, impaired speech, and behavioral abnormalities, 619475
SPTBN2	100%	100%	100%	100%	99.2%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SPTBN4	100%	100%	100%	100%	99%	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness, 617519
SRCAP	100%	100%	100%	100%	99.1%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRD5A3	100%	100%	100%	100%	99.8%	Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SRP54	96.3%	96.3%	100%	100%	99.9%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRPK3	100%	100%	98.6%	86.5%	67.9%	Intellectual developmental disorder, X-linked 114, 301134

SRPX2	100%	100%	98.7%	88.3%	67.1%	?Rolandic epilepsy, impaired intellectual development, and speech dyspraxia, 300643
SRRM1	100%	100%	100%	100%	99.6%	
SRRM2	100%	100%	100%	99.9%	98.5%	Intellectual developmental disorder, autosomal dominant 72, 620439
SRRM4	100%	100%	100%	99.9%	99.3%	
SRSF1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with dysmorphic facies and behavioral abnormalities, 620489
SSPOP						
SSR4	100%	100%	97.5%	84.4%	64.1%	Congenital disorder of glycosylation, type Iy, 300934
ST3GAL3	89.8%	88.2%	100%	100%	99.5%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.4%	98.4%	100%	100%	99.4%	Salt and pepper developmental regression syndrome, 609056
STAG1	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 47, 617635
STAG2	100%	100%	99.1%	90.7%	72.9%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261

STEEP1	100%	100%	98.7%	89.1%	70.1%	?Intellectual developmental disorder, X-linked 107, 301013
STIL	100%	100%	100%	100%	99.9%	Microcephaly 7, primary, autosomal recessive, 612703
STRA6	100%	100%	100%	100%	99.5%	Microphthalmia, syndromic 9, 601186;Microphthalmia, isolated, with coloboma 8, 601186
STRADA	100%	100%	100%	100%	99.6%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STT3A	100%	99.6%	100%	100%	99.8%	Congenital disorder of glycosylation, type Iw, autosomal dominant, 619714;Congenital disorder of glycosylation, type Iw, autosomal recessive, 615596
STT3B	100%	100%	100%	99.8%	99.2%	Congenital disorder of glycosylation, type Ix, 615597
STX1A	100%	99%	100%	99.9%	99.2%	
STX1B	100%	100%	100%	100%	98.6%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 4, 612164
SUCLA2	96.5%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

SUCLG1	100%	100%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400
SUFU	100%	100%	100%	100%	99.3%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SUMF1	100%	100%	100%	100%	99.4%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	99.9%	99.3%	Sulfite oxidase deficiency, 272300
SUPT16H	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with dysmorphic facies and thin corpus callosum, 619480
SUPT4H1	100%	100%	100%	100%	99.8%	
SURF1	100%	100%	100%	100%	99.2%	Charcot-Marie-Tooth disease, type 4K, 616684;Mitochondrial complex IV deficiency, nuclear type 1, 220110
SUZ12	95.6%	95.6%	100%	100%	99.6%	Imagawa-Matsumoto syndrome, 618786
SVBP	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569
SYN1	100%	100%	97.2%	85.1%	65.2%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNCRIP	92.9%	92.9%	100%	100%	99.8%	

SYNGAP1	100%	100%	100%	99.9%	98.9%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100%	100%	100%	100%	99.7%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYP	100%	100%	98.3%	85.3%	64.6%	Intellectual developmental disorder, X-linked 96, 300802
SYT1	96%	96%	100%	100%	99.8%	Baker-Gordon syndrome, 618218
SZT2	100%	100%	100%	99.9%	99.6%	Developmental and epileptic encephalopathy 18, 615476
TACO1	100%	100%	100%	100%	99.6%	Mitochondrial complex IV deficiency, nuclear type 8, 619052
TAF1	98.7%	98.7%	98.5%	87.8%	68.8%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TAF13	95.2%	82.4%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 60, 617432
TAF1C	100%	99.4%	100%	100%	99.2%	
TAF2	96.3%	96.3%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 40, 615599
TAF4	91.9%	84.9%	100%	98.7%	93.1%	Intellectual developmental disorder, autosomal dominant 73, 620450

TAF6	100%	100%	100%	100%	99.2%	Alazami-Yuan syndrome, 617126
TAF8	85.3%	85.3%	100%	100%	99.2%	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972
TANC2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder with autistic features and language delay, with or without seizures, 618906
TANGO2	100%	100%	100%	100%	99.9%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TAOK1	100%	100%	100%	100%	99.7%	Developmental delay with or without intellectual impairment or behavioral abnormalities, 619575
TAOK2	100%	100%	100%	99.9%	99.1%	
TARS2	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 21, 615918
TASP1	93.2%	93.2%	100%	100%	99.9%	Suleiman-El-Hattab syndrome, 618950
TAT	100%	100%	100%	100%	99.9%	Tyrosinemia, type II, 276600
TBC1D20	92.2%	92.2%	100%	100%	99.5%	Warburg micro syndrome 4, 615663
TBC1D23	96.3%	96.3%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 11, 617695

TBC1D24	100%	100%	100%	100%	99.6%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBC1D2B	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with seizures and gingival overgrowth, 619323
TBC1D7	88.9%	88.9%	100%	100%	99.7%	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000
TBCB	77.8%	77.5%	100%	99.9%	98.8%	Neurodevelopmental disorder with behavioral abnormalities and childhood onset spastic paraplegia, 621382
TBCD	91.3%	91%	100%	99.9%	99.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	97.7%	96.6%	100%	100%	99.7%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207

TBCK	100%	100%	100%	100%	99.9%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900
TBL1XR1	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 41, 616944;Pierpont syndrome, 602342
TBP	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia 17, 607136;{Parkinson disease, susceptibility to}, 168600
TBR1	100%	100%	100%	99.9%	98.8%	Intellectual developmental disorder with autism and speech delay, 606053
TBX1	98.4%	95.8%	100%	99.9%	97.2%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TCEAL1	100%	100%	98%	86.9%	70.4%	Hijazi-Reis syndrome, 301094
TCF20	100%	100%	100%	100%	99.6%	Developmental delay with variable intellectual impairment and behavioral abnormalities, 618430
TCF4	97.1%	97.1%	100%	100%	99.9%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TCF7L2	100%	100%	100%	99.9%	99.2%	{Diabetes mellitus, type 2, susceptibility to}, 125853
TCN2	92.6%	88.3%	100%	100%	99.7%	Transcobalamin II deficiency, 275350

TCP1	100%	100%	100%	100%	99.6%	Intellectual developmental disorder with polymicrogyria and seizures, 621021
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.9%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TDP2	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100%	100%	100%	100%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TECR	87.3%	87.3%	100%	99.8%	99.1%	Intellectual developmental disorder, autosomal recessive 14, 614020
TEFM	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 58, 620451
TELO2	94.4%	94.3%	100%	100%	99.5%	You-Hoover-Fong syndrome, 616954
TENM3	99.6%	99.6%	100%	100%	99.6%	Microphthalmia, syndromic 15, 615145;?Microphthalmia/coloboma 9, 615145
TET3	100%	100%	100%	99.9%	98.9%	Beck-Fahrner syndrome, 618798
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
TFE3	100%	99.9%	98.1%	86.7%	66.5%	Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854

TGDS	100%	100%	100%	100%	99.9%	Catel-Manzke syndrome, 616145
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGIF1	100%	100%	100%	100%	99.7%	Holoprosencephaly 4, 142946
TH	100%	100%	100%	100%	99.4%	Segawa syndrome, recessive, 605407
THG1L	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 28, 618800
THOC2	100%	100%	99.4%	90.4%	71.9%	Arthrogryposis multiplex congenita 7, X-linked, 301127;Intellectual developmental disorder, X-linked syndromic, Kumar type, 300957
THOC6	100%	100%	100%	100%	99.2%	Beaulieu-Boycott-Innes syndrome, 613680
THRB	100%	100%	100%	99.9%	99.6%	Thyroid hormone resistance, autosomal recessive, 274300;Thyroid hormone resistance, 188570;Thyroid hormone resistance, selective pituitary, 145650
THUMPD1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with speech delay and variable ocular anomalies, 619989
TIAM1	98.3%	98.3%	100%	100%	99.6%	Neurodevelopmental disorder with language delay and seizures, 619908
TIMM50	100%	100%	100%	99.9%	99.2%	3-methylglutaconic aciduria, type IX, 617698

TIMM8A	100%	100%	98.7%	88.1%	68%	Mohr-Tranebjaerg syndrome, 304700
TINF2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TKFC	100%	100%	100%	100%	99.5%	Triokinase and FMN cyclase deficiency syndrome, 618805
TKT	89.3%	89%	100%	100%	99.6%	Short stature, developmental delay, and congenital heart defects, 617044
TLK2	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 57, 618050
TM2D3	100%	100%	100%	100%	99.8%	Neurocardiorenal malformation syndrome, 621379
TMCO1	72.3%	72.3%	100%	99.9%	99.5%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM106B	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM107	100%	100%	100%	100%	99.1%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM147	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with facial dysmorphism, absent language, and pseudo-Pelger-Huet anomaly, 620075
TMEM163	93.9%	93.9%	100%	99.9%	99.1%	Leukodystrophy, hypomyelinating, 25, 620243

TMEM165	88.9%	88.9%	100%	100%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM167A	100%	100%	100%	100%	99.9%	
TMEM184B	100%	100%	100%	100%	99.5%	
TMEM216	100%	100%	100%	100%	99.9%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM218	100%	100%	100%	99.9%	99.3%	Joubert syndrome 39, 619562
TMEM222	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with motor and speech delay and behavioral abnormalities, 619470
TMEM231	93.5%	93.5%	100%	100%	99.4%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397
TMEM237	98.2%	98.2%	100%	100%	99.8%	Joubert syndrome 14, 614424
TMEM240	100%	100%	100%	99.9%	98.5%	Spinocerebellar ataxia 21, 607454
TMEM63A	99.8%	98.2%	100%	100%	99.6%	Leukodystrophy, hypomyelinating, 19, transient infantile, 618688
TMEM63B	100%	100%	100%	99.9%	99.3%	Developmental and epileptic encephalopathy 118, 621250
TMEM63C	100%	100%	100%	100%	99.5%	Spastic paraplegia 87, autosomal recessive, 619966

TMEM67	96.1%	96.1%	100%	100%	99.8%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TMEM70	100%	100%	100%	100%	99.8%	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052
TMEM94	100%	100%	100%	100%	99.4%	Intellectual developmental disorder with cardiac defects and dysmorphic facies, 618316
TMLHE	94.8%	94.8%	98.7%	91.1%	74.5%	{Autism, susceptibility to, X-linked 6}, 300872
TMTC3	100%	100%	100%	100%	99.8%	Lissencephaly 8, 617255
TMX2	100%	99%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730
TNIK	98.6%	98.6%	100%	100%	99.6%	Intellectual developmental disorder, autosomal recessive 54, 617028
TNPO2	100%	100%	100%	100%	98.7%	Intellectual developmental disorder with hypotonia, impaired speech, and dysmorphic facies, 619556
TNR	100%	100%	100%	100%	99.5%	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus, 619653

TNRC6B	100%	100%	100%	100%	99.6%	Global developmental delay with speech and behavioral abnormalities, 619243
TOE1	100%	100%	100%	99.9%	98.6%	Pontocerebellar hypoplasia, type 7, 614969
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOMM70	100%	100%	100%	100%	99.9%	
TOR1A	95.6%	91.3%	100%	100%	99.5%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TP53RK	100%	100%	100%	100%	99.7%	Galloway-Mowat syndrome 4, 617730
TP73	100%	100%	100%	99.9%	99.1%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TPI1	100%	100%	100%	100%	99.6%	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512
TPO	100%	100%	100%	100%	99.5%	Thyroid dyshormonogenesis 2A, 274500
TPP1	100%	100%	100%	100%	99.6%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TPP2	100%	100%	100%	100%	99.8%	Immunodeficiency 78 with autoimmunity and developmental delay, 619220
TPRKB	89.7%	81.9%	100%	100%	99.3%	Galloway-Mowat syndrome 5, 617731
TRA2B	100%	100%	100%	100%	99.6%	
TRAF7	100%	100%	100%	99.9%	99%	Cardiac, facial, and digital anomalies with developmental delay, 618164

TRAIP	100%	100%	100%	100%	99.4%	Seckel syndrome 9, 616777
TRAK1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 68, 618201
TRAPPC11	100%	100%	100%	100%	99.8%	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356
TRAPPC12	87.1%	86.3%	100%	100%	99.3%	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669
TRAPPC2L	100%	100%	100%	100%	99.6%	Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis, 618331
TRAPPC4	100%	100%	100%	99.9%	98.9%	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741
TRAPPC6B	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy, 617862
TRAPPC9	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 13, 613192
TREX1	100%	100%	100%	99.9%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448

TRH	100%	100%	100%	100%	99.2%	Thyrotropin-releasing hormone deficiency, 275120
TRIM32	100%	100%	100%	100%	99.2%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM71	100%	100%	100%	99.9%	98.2%	Hydrocephalus, congenital, 4, 618667
TRIM8	100%	100%	100%	99.8%	98.3%	Focal segmental glomerulosclerosis and neurodevelopmental syndrome, 619428
TRIO	99.3%	99.1%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 44, with microcephaly, 617061;Intellectual developmental disorder, autosomal dominant 63, with macrocephaly, 618825
TRIP12	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 49, 617752
TRIT1	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 35, 617873
TRMT1	100%	100%	100%	100%	99.3%	Intellectual developmental disorder, autosomal recessive 68, 618302
TRMT10A	100%	100%	100%	100%	99.7%	Microcephaly, short stature, and impaired glucose metabolism 1, 616033
TRMT5	100%	100%	100%	100%	99.8%	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay, 616539

TRNT1	92%	91.9%	100%	100%	99.8%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959
TRPM3	97.8%	97.8%	100%	100%	99.7%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TRPM7	100%	100%	100%	100%	99.8%	{Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to}, 105500
TRRAP	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TSC1	100%	100%	100%	100%	99.7%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	100%	100%	100%	100%	99.4%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSEN15	100%	100%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 2F, 617026

TSEN2	88.4%	88.4%	100%	100%	99.8%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100%	100%	100%	100%	99.3%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TSFM	93.6%	91.4%	100%	100%	99.6%	Combined oxidative phosphorylation deficiency 3, 610505
TSHB	100%	100%	100%	100%	99.8%	Hypothyroidism, congenital, nongoitrous 4, 275100
TSPAN7	100%	100%	99%	89%	69.5%	Intellectual developmental disorder, X-linked 58, 300210
TSPOAP1	100%	100%	100%	99.9%	99.4%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456
TTC19	100%	100%	100%	100%	99.8%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTC5	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with cerebral atrophy and variable facial dysmorphism, 619244
TTC8	91.4%	91.4%	100%	100%	99.8%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TTI1	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with microcephaly and movement abnormalities, 620445
TTI2	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 39, 615541

TUBA1A	100%	100%	100%	100%	99.7%	Lissencephaly 3, 611603
TUBA8	95.5%	95.3%	100%	100%	99.4%	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840
TUBB	99.9%	99.3%	100%	100%	99.5%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB2A	100%	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	100%	100%	99.9%	98.9%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB3	100%	100%	100%	99.9%	99.1%	Fibrosis of extraocular muscles, congenital, 3A, 600638;Cortical dysplasia, complex, with other brain malformations 1, 614039
TUBB4A	99.8%	97.9%	100%	100%	99.2%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100%	100%	100%	100%	99.5%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TUBGCP2	97%	97%	100%	99.9%	99.3%	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures, 618737

TUBGCP4	100%	100%	99.8%	99.2%	97.7%	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335
TUBGCP6	100%	100%	100%	100%	99.2%	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270
TUSC3	93.3%	93.3%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 7, 611093
TWIST1	100%	100%	100%	99.9%	99%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TWNK	100%	100%	100%	100%	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%	100%	100%	99.8%	98.5%	Developmental delay, dysmorphic facies, and brain anomalies, 620535
UBA2	100%	100%	100%	100%	99.8%	ACCES syndrome, 619959
UBA5	94.1%	92.7%	100%	100%	99.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132

UBAP2L	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with impaired language, behavioral abnormalities, and dysmorphic facies, 620494
UBE2A	95.9%	92.5%	98.8%	89.6%	70.2%	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBE3A	100%	100%	100%	100%	99.8%	Angelman syndrome, 105830
UBE3B	100%	100%	100%	100%	99.6%	Kaufman oculocerebrofacial syndrome, 244450
UBE3C	94.8%	94.8%	100%	100%	99.6%	Neurodevelopmental disorder with absent speech and movement and behavioral abnormalities, 620270
UBE4A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with hypotonia and gross motor and speech delay, 619639
UBR1	98%	98%	100%	100%	99.7%	Johanson-Blizzard syndrome, 243800
UBR5	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with speech delay and behavioral abnormalities, 621372
UBR7	100%	100%	100%	100%	99.6%	Li-Campeau syndrome, 619189
UBTF	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UFC1	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with spasticity and poor growth, 618076
UFM1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 14, 617899

UFSP2	100%	100%	100%	100%	99.8%	?Hip dysplasia, Beukes type, 142669;Spondyloepimetaphyseal dysplasia, Di Rocco type, 617974;Developmental and epileptic encephalopathy 106, 620028
UGDH	98.8%	95.4%	100%	100%	99.7%	Developmental and epileptic encephalopathy 84, 618792
UGGT1	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type IICC, 621381
UGP2	93.2%	91.3%	100%	100%	99.7%	Developmental and epileptic encephalopathy 83, 618744
UNC13A	100%	100%	100%	100%	99.2%	
UNC45A	100%	100%	100%	100%	99.7%	Osteootohepatoenteric syndrome, 619377
UNC79	100%	100%	100%	100%	99.7%	
UNC80	100%	100%	100%	100%	99.6%	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801
UPB1	95.4%	92.1%	100%	100%	99.8%	Beta-ureidopropionase deficiency, 613161
UPF1	99.2%	96.5%	100%	100%	99.4%	
UPF3B	92.5%	92.5%	98.8%	88%	69.9%	Intellectual developmental disorder, X-linked syndromic 14, 300676
UROC1	96.6%	96.6%	100%	100%	99.4%	?Urocanase deficiency, 276880
USP14	100%	100%	100%	100%	99.8%	
USP27X	100%	100%	98.6%	89.7%	70.5%	Intellectual developmental disorder, X-linked 105, 300984

USP7	100%	100%	100%	100%	99.4%	Hao-Fountain syndrome, 616863
USP9X	100%	100%	99.2%	91%	71.7%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
VAMP1	100%	100%	100%	100%	99.3%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VAMP2	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements, 618760
VARS1	100%	100%	100%	99.9%	99.2%	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802
VARS2	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 20, 615917
VCP	100%	99.9%	100%	100%	99.4%	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
VIPAS39	100%	98.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

VLDLR	100%	100%	100%	100%	99.7%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VMA22	100%	99.1%	100%	99.9%	98.7%	Congenital disorder of glycosylation, type IIo, 616828
VPS11	100%	100%	100%	100%	99.5%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS16	100%	100%	100%	100%	99.6%	Dystonia 30, 619291
VPS35L	95.1%	93.3%	100%	100%	99.5%	Ritscher-Schinzel syndrome 3, 619135
VPS37A	92.3%	92.3%	100%	100%	99.7%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	98.7%	97.7%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS4A	100%	100%	100%	99.9%	98.1%	CIMDAG syndrome, 619273
VPS50	97%	95.5%	100%	100%	99.9%	Neurodevelopmental disorder with microcephaly, seizures, and neonatal cholestasis, 619685
VPS53	83.4%	82.4%	100%	100%	99.6%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	100%	99.2%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
VWA3B	100%	100%	100%	100%	99.5%	?Spinocerebellar ataxia, autosomal recessive 22, 616948

WAC	94%	94%	100%	100%	99.7%	Desanto-Shinawi syndrome, 616708
WAPL	97.6%	97.6%	100%	100%	99.7%	
WARS1	91.8%	91.8%	100%	100%	99.1%	Neuronopathy, distal hereditary motor, autosomal dominant 9, 617721;Neurodevelop mental disorder with microcephaly and speech delay, with or without brain abnormalities, 620317
WARS2	93.8%	93.8%	100%	100%	99.9%	Parkinsonism-dystonia 3, childhood-onset, 619738;Neurodevelop mental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710
WASF1	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with absent language and variable seizures, 618707
WASHC4	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal recessive 43, 615817
WASHC5	100%	100%	100%	100%	99.9%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563
WBP4	100%	100%	100%	99.9%	99.1%	Neurodevelopemental disorder with hypotonia, feeding difficulties, facial dysmorphism, and brain abnormalities, 620852
WDFY3	100%	100%	100%	100%	99.8%	?Microcephaly 18, primary, autosomal dominant, 617520

WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	100%	100%	100%	100%	99.8%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR13	100%	100%	98.6%	87.8%	67.9%	
WDR26	93.8%	93.8%	100%	100%	99.6%	Skraban-Deardorff syndrome, 617616
WDR37	90.5%	90%	100%	99.9%	99.5%	Neurooculocardiogenitourinary syndrome, 618652
WDR4	100%	100%	100%	100%	99.1%	Galloway-Mowat syndrome 6, 618347;Microcephaly, growth deficiency, seizures, and brain malformations, 618346
WDR44	91.1%	91.1%	99%	90.1%	70.7%	
WDR45	99.1%	98.2%	98.6%	86.8%	66.4%	Neurodegeneration with brain iron accumulation 5, 300894
WDR45B	89.6%	89.6%	100%	100%	99.2%	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977
WDR47	100%	100%	100%	100%	99.8%	
WDR5	93.9%	93.9%	100%	100%	99.4%	
WDR62	98.8%	98.8%	100%	100%	99.6%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317

WDR73	100%	100%	100%	99.9%	99.6%	Galloway-Mowat syndrome 1, 251300
WDR81	100%	100%	100%	100%	99.5%	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185;Hydrocephalus, congenital, 3, with brain anomalies, 617967
WDR83OS	100%	100%	100%	99.8%	99.4%	Neurodevelopmental disorder with variable familial hypercholanemia, 621016
WFS1	91.5%	91.2%	100%	100%	99.5%	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%	100%	100%	100%	99.5%	?Intellectual developmental disorder with short stature and variable skeletal anomalies, 618453
WLS	100%	100%	100%	100%	99.8%	Zaki syndrome, 619648
WNK3	100%	100%	99.3%	90.6%	72%	Prieto syndrome, 309610
WNT1	100%	100%	100%	100%	99.6%	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221;Osteogenesis imperfecta, type XV, 615220
WSB2	100%	100%	100%	100%	99.6%	

WWOX	100%	100%	100%	100%	99.7%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XPA	100%	100%	100%	100%	99.8%	Xeroderma pigmentosum, group A, 278700
XPO1	100%	100%	100%	100%	99.9%	
XPOT	89.9%	89.4%	100%	99.9%	99.6%	
XRCC4	100%	100%	100%	100%	99.9%	Short stature, microcephaly, and endocrine dysfunction, 616541
XYLT1	100%	100%	100%	99.9%	99.1%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
YARS1	100%	99.3%	100%	100%	99.7%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418;Charcot-Marie-Tooth disease, dominant intermediate C, 608323
YIF1B	90%	90%	100%	99.9%	99.2%	Kaya-Barakat-Masson syndrome, 619125
YIPF5	100%	100%	100%	100%	99.9%	Microcephaly, epilepsy, and diabetes syndrome 2, 619278
YME1L1	96.7%	96.7%	100%	100%	99.8%	?Optic atrophy 11, 617302
YWHAE	100%	100%	100%	100%	99.8%	
YWHAG	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 56, 617665

YWHAZ	100%	100%	100%	100%	99.7%	
YY1	100%	100%	100%	99.4%	96.4%	Gabriele-de Vries syndrome, 617557
YY1AP1	100%	100%	100%	100%	99.5%	Grange syndrome, 602531
ZBTB11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 69, 618383
ZBTB16	87.8%	87.8%	100%	100%	99.6%	Leukemia, acute promyelocytic, PL2F/RARA type
ZBTB18	93.2%	93.2%	100%	100%	99.3%	Intellectual developmental disorder, autosomal dominant 22, 612337
ZBTB20	100%	100%	100%	99.9%	98.9%	Primrose syndrome, 259050
ZBTB24	100%	100%	100%	100%	99.5%	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069
ZBTB47	99.6%	99.5%	100%	99.9%	98.2%	
ZBTB7A	95.4%	95.4%	100%	99.9%	97.3%	Macrocephaly, neurodevelopmental delay, lymphoid hyperplasia, and persistent fetal hemoglobin, 619769
ZC3H14	100%	100%	100%	99.9%	99.7%	Intellectual developmental disorder, autosomal recessive 56, 617125
ZC4H2	100%	100%	98.5%	88.7%	67.5%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041

ZDHHC9	100%	100%	98.9%	88.4%	70.2%	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799
ZEB2	100%	100%	100%	99.9%	99.3%	Mowat-Wilson syndrome, 235730
ZFHX3	100%	100%	100%	100%	99.3%	Prostate cancer, somatic, 176807;{Atrial fibrillation 8, susceptibility to}, 613055;Spinocerebellar ataxia 4, 600223
ZFHX4	100%	100%	100%	100%	99.7%	
ZFTRAF1	75.2%	72.2%	100%	99.6%	97.2%	
ZFX	100%	100%	98.9%	90.6%	72.5%	Intellectual developmental disorder, X-linked syndromic 37, 301118
ZFYVE26	100%	100%	100%	100%	99.6%	Spastic paraplegia 15, autosomal recessive, 270700
ZIC1	100%	100%	100%	100%	99.2%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	100%	100%	99.7%	96.4%	Holoprosencephaly 5, 609637
ZMIZ1	100%	99.6%	100%	99.8%	98.9%	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies, 618659
ZMYM2	100%	100%	100%	100%	99.8%	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities, 619522

ZMYM3	100%	100%	98.2%	86.7%	66.8%	Intellectual developmental disorder, X-linked 112, 301111
ZMYND11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal dominant 30, 616083
ZMYND8	98%	98%	100%	100%	99.6%	
ZNF142	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with impaired speech and hyperkinetic movements, 618425
ZNF148	94.9%	94.9%	100%	100%	99.8%	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260
ZNF292	99.4%	99.4%	100%	100%	99.8%	Intellectual developmental disorder, autosomal dominant 64, 619188
ZNF335	100%	100%	100%	100%	99.5%	Microcephaly 10, primary, autosomal recessive, 615095
ZNF407	100%	100%	100%	100%	99.6%	SIMHA syndrome, 619557
ZNF41	100%	100%	99.1%	89.7%	71.4%	
ZNF462	100%	100%	100%	100%	99.5%	Weiss-Kruszka syndrome, 618619
ZNF526	100%	100%	100%	100%	99.4%	Dentici-Novelli neurodevelopmental syndrome, 619877
ZNF668	100%	100%	100%	100%	99.1%	Neurodevelopmental disorder with poor growth, large ears, and dysmorphic facies, 620194
ZNF699	100%	100%	100%	100%	99.6%	DEGCAGS syndrome, 619488

ZNF711	100%	100%	99.2%	91.7%	73.5%	Intellectual developmental disorder, X-linked 97, 300803
ZNF865	100%	100%	100%	100%	98.9%	
ZNFX1	100%	100%	100%	100%	99.7%	Immunodeficiency 91 and hyperinflammation, 619644
ZNHIT3	78.5%	76.2%	100%	100%	99.6%	PEHO syndrome, 260565
ZNRF3	98.5%	97%	100%	99.9%	99%	
ZRSR2	84.4%	84.4%	98.8%	88.9%	70.3%	Orofaciodigital syndrome XXI, 301132
ZSCAN10	100%	100%	100%	100%	99.3%	Otofacial neurodevelopmental syndrome, 620910
ZSWIM6	97.9%	96.4%	99.5%	98.2%	96.1%	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 mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors