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

MOVEMENT DISORDERS PANEL DG-5.1.0 (452 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*
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCB7	100%	100%	99.6%	92.7%	74.7%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ABHD12	100%	100%	100%	100%	99.6%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ABHD16A	97.5%	97.5%	100%	100%	99.4%	Spastic paraplegia 86, autosomal recessive, 619735
ACO2	90.7%	86.8%	100%	100%	99.7%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559

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

ACTB	100%	100%	100%	99.9%	99.4%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ADAR	100%	100%	100%	100%	99.8%	Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
ADCY5	97.5%	96.9%	100%	99.9%	99.1%	Dyskinesia with orofacial involvement, autosomal dominant, 606703;Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651;Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADD3	97.6%	97.4%	100%	100%	99.6%	Cerebral palsy, spastic quadriplegic, 3, 617008
ADGRG1	100%	100%	100%	100%	99.3%	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752;Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854

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

ADPRS	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	94.2%	92.2%	100%	100%	99.5%	Spastic ataxia 5, autosomal recessive, 614487;Optic atrophy 12, 618977;Spinocerebellar ataxia 28, 610246
AGA	91.6%	91.6%	100%	100%	99.5%	Aspartylglucosaminuria, 208400
AGTPBP1	96.5%	96.5%	100%	100%	99.7%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIFM1	100%	100%	99.3%	90.3%	72.6%	Combined oxidative phosphorylation deficiency 6, 300816;Cowchock syndrome, 310490;Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232;Deafness, X-linked 5, 300614
AIMP1	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 3, 260600
ALDH18A1	100%	100%	100%	100%	99.6%	Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603
ALDH3A2	93.5%	93.5%	100%	100%	99.7%	Sjogren-Larsson syndrome, 270200

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

ALDH5A1	100%	100%	100%	100%	99.5%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALS2	97%	96.3%	100%	100%	99.8%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100
AMFR	100%	100%	100%	100%	99.6%	Spastic paraplegia 89, autosomal recessive, 620379
AMPD2	100%	100%	100%	99.9%	99.6%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686
ANO10	94.9%	94.9%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	100%	100%	100%	100%	99.8%	Dystonia 24, 615034
AOPEP	100%	100%	100%	100%	99.5%	Dystonia 31, 619565
AP4B1	100%	100%	100%	100%	99.8%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100%	100%	100%	100%	99.9%	Stuttering, familial persistent, 1, 184450;Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100%	100%	100%	100%	99.7%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	79.1%	79.1%	100%	99.9%	99.4%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	91.2%	91%	100%	100%	99.5%	Spastic paraplegia 48, autosomal recessive, 613647

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

APTX	100%	100%	100%	100%	99.7%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	93%	93%	100%	100%	99.8%	Argininemia, 207800
ARSA	100%	100%	100%	99.9%	99%	Metachromatic leukodystrophy, 250100
ARX	99.9%	98.4%	96.3%	80.9%	59.5%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419
ASPA	100%	100%	100%	100%	99.8%	Canavan disease, 271900
ATCAY	100%	100%	100%	100%	99.2%	Ataxia, cerebellar, Cayman type, 601238
ATG7	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 31, 619422
ATL1	100%	100%	100%	100%	99.7%	Spastic paraplegia 3A, autosomal dominant, 182600;Neuropathy, hereditary sensory, type ID, 613708
ATM	100%	100%	100%	100%	99.7%	Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic

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

ATN1	98.2%	98.2%	100%	100%	98.7%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP13A2	100%	100%	100%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A2	100%	100%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481
ATP1A3	100%	100%	100%	100%	99.2%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP2B3	100%	100%	98.7%	88.2%	69%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	100%	100%	100%	100%	99.6%	Wilson disease, 277900
ATP8A2	100%	100%	100%	100%	99.6%	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268

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

B4GALNT1	100%	100%	100%	100%	99.5%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	91.5%	86.7%	98.7%	88.7%	70.4%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	86.3%	86.3%	100%	99.9%	99.3%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89.9%	87.7%	100%	99.9%	99.3%	Maple syrup urine disease, type Ib, 620698
BCL11B	100%	99.8%	100%	99.9%	98%	Immunodeficiency 49, severe combined, 617237;Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BRAT1	97.3%	96.5%	100%	99.9%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BSCL2	100%	100%	100%	100%	99.6%	Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	94.2%	94.2%	100%	100%	99.9%	Biotinidase deficiency, 253260

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

C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
CA8	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 34, 613227
CACNA1A	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500
CACNA1E	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 69, 618285
CACNA1G	100%	100%	100%	99.9%	99.4%	Spinocerebellar ataxia 42, 616795;Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CAMTA1	100%	100%	100%	100%	99.2%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CAPN1	100%	100%	100%	100%	99.2%	Spastic paraplegia 76, autosomal recessive, 616907
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285

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

CCT5	100%	100%	100%	100%	99.5%	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHMP1A	100%	100%	100%	99.9%	99.2%	Pontocerebellar hypoplasia, type 8, 614961
CLCN2	100%	99.1%	100%	100%	99.6%	Leukoencephalopathy with ataxia, 615651;Hyperaldosteronism, familial, type II, 605635;{Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628;{Epilepsy, juvenile absence, susceptibility to, 2}, 607628;{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628
CLCN4	100%	100%	98.5%	88.6%	71.1%	Raynaud-Claes syndrome, 300114
CLCN6	97.7%	97.7%	100%	100%	99.6%	Ceroid lipofuscinosis, neuronal, 15, 619173
CLN5	83%	83%	100%	100%	99.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	95.1%	95.1%	100%	100%	99.4%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLP1	100%	100%	100%	100%	99.5%	Pontocerebellar hypoplasia, type 10, 615803

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

CLPB	98.2%	98.2%	100%	100%	99.7%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
COASY	100%	100%	100%	99.8%	99.1%	Pontocerebellar hypoplasia, type 12, 618266;Neurodegeneration with brain iron accumulation 6, 615643
COL4A1	100%	100%	100%	100%	99.7%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	98.6%	98.5%	100%	100%	99.3%	Brain small vessel disease 2A, autosomal dominant, 614483;{Hemorrhage, intracerebral, susceptibility to}, 614519;Brain small vessel disease 2B, autosomal recessive, 621414
COL6A1	100%	100%	100%	100%	99.4%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810

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

COL6A2	100%	99.8%	100%	99.9%	99.2%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	100%	99.7%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COQ2	96.3%	96.3%	100%	100%	99.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	96.3%	96.3%	100%	100%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ8A	89.1%	87.2%	100%	99.9%	99.5%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	91.9%	91.9%	100%	99.9%	99.2%	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	100%	100%	100%	100%	99.8%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
CP	100%	100%	100%	100%	99.9%	Aceruloplasminemia, 604290
CSF1R	97.3%	97.3%	100%	100%	99.6%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSTB	100%	100%	100%	100%	99.4%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800

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

CTBP1	100%	99.6%	100%	99.8%	99.3%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTC1	100%	100%	100%	100%	99.4%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
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
CTSD	86%	86%	100%	100%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	100%	99.9%	99.4%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CWF19L1	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYP27A1	100%	100%	100%	100%	99.5%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100%	100%	100%	99.9%	98.8%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	100%	100%	100%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800;Bile acid synthesis defect, congenital, 3, 613812

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

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;Charcot-Marie-Tooth disease, axonal, type 2LL, 621485
DBT	100%	100%	100%	100%	99.9%	Maple syrup urine disease, type II, 620699
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	99.7%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DCTN1	100%	100%	100%	100%	99%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641
DDC	100%	100%	100%	100%	99.7%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100%	100%	100%	100%	99.3%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	100%	100%	100%	99.7%	Spastic paraplegia 54, autosomal recessive, 615033

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

DEGS1	100%	100%	100%	100%	99.8%	Leukodystrophy, hypomyelinating, 18, 618404
DHDDS	74.4%	74.4%	100%	100%	99.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 621567;Retinitis pigmentosa 59, 613861
DHPS	96.4%	93.1%	100%	99.9%	99%	Neurodevelopmental disorder with seizures and speech and walking impairment, 618480
DHX30	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with variable motor and speech impairment, 617804
DLAT	100%	100%	100%	100%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100%	100%	100%	100%	99.8%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAJC12	100%	100%	100%	100%	99.7%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC3	92.1%	92.1%	100%	100%	99.8%	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAL4	100%	100%	100%	99.8%	98%	?Mirror movements 3, 616059
DNM1L	100%	100%	100%	100%	99.9%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388

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

DNMT1	100%	99.3%	100%	100%	99.4%	Neuropathy, hereditary sensory, type IE, 614116;Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DPYS	100%	100%	100%	100%	99.5%	Dihydropyrimidinuria, 222748
DSTYK	100%	100%	100%	100%	99.5%	Spastic paraplegia 23, autosomal recessive, 270750;Congenital anomalies of kidney and urinary tract 1, 610805
DTYMK	91.4%	84.2%	100%	100%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
EBF3	100%	100%	100%	99.9%	99.2%	Hypotonia, ataxia, and delayed development syndrome, 617330
ECHS1	71.2%	69%	100%	100%	99.3%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2AK1	95%	94.1%	100%	100%	99.7%	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878
EIF2AK2	100%	100%	100%	100%	99.8%	Leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome, 618877;Dystonia 33, 619687
EIF2B1	93.5%	90.6%	100%	100%	99.8%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100%	100%	100%	99.9%	99.2%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312

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

EIF2B3	100%	100%	100%	100%	99.5%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	100%	99.9%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	100%	99.6%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
ELOVL1	100%	100%	100%	100%	99.9%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
ELOVL5	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia 38, 615957
ENTPD1	100%	100%	100%	100%	99.6%	Spastic paraplegia 64, autosomal recessive, 615683
ERCC2	96.3%	94.6%	100%	100%	99.3%	Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756

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

ERCC4	100%	100%	100%	100%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERLIN1	100%	100%	100%	100%	99.8%	Spastic paraplegia 62, autosomal recessive, 615681
ERLIN2	100%	100%	100%	100%	99.5%	Spastic paraplegia 18A, autosomal dominant, 620512;Spastic paraplegia 18B, autosomal recessive, 611225
ETHE1	77.1%	77.1%	100%	100%	99.7%	Ethylmalonic encephalopathy, 602473
EXOSC3	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	100%	98.6%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100%	100%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 1D, 618065
FA2H	100%	100%	100%	100%	99.7%	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	91%	90.9%	100%	100%	99.9%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154;Cataracts, spastic paraparesis, and speech delay, 619338

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

FARS2	100%	100%	100%	100%	99.5%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FBXO7	99.1%	98.1%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FGF14	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 27A, 193003;Spinocerebellar ataxia 27B, late-onset, 620174
FICD	100%	100%	100%	100%	99.2%	Spastic paraplegia 92, autosomal recessive, 620911
FLVCR1	95%	94.6%	100%	100%	99.7%	Retinopathy-sensory neuropathy syndrome, 609033;Neurodevelop mental disorder with microcephaly, absent speech, and hypotonia, 621060
FOLR1	100%	100%	100%	100%	99.5%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD5	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094
FRMD7	100%	100%	99.5%	93.2%	75.5%	Nystagmus, infantile periodic alternating, X-linked, 310700;Nystagmus 1, congenital, X-linked, 310700
FTH1	100%	100%	100%	100%	99.7%	Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromat osis, type 5, 615517

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

FTL	100%	100%	100%	100%	99.3%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
FXN	100%	100%	100%	100%	98.9%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
GALC	100%	100%	100%	100%	99.6%	Krabbe disease, 245200
GAMT	97%	93.2%	100%	99.8%	98.8%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100%	100%	100%	99.9%	99.5%	Giant axonal neuropathy-1, 256850
GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	100%	100%	100%	99.9%	99.4%	Spastic paraplegia 46, autosomal recessive, 614409
GBE1	100%	100%	100%	100%	99.7%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GCDH	79.8%	79.8%	100%	99.9%	99.5%	Glutaricaciduria, type I, 231670

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

GCH1	100%	100%	100%	100%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
GDAP2	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GEMIN5	100%	100%	100%	99.9%	99.6%	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333
GFAP	100%	100%	100%	99.9%	99.4%	Alexander disease, 203450
GJB1	100%	100%	97.9%	84.9%	64.7%	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800
GJC2	100%	99.4%	100%	99.9%	98.3%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GLB1	100%	100%	100%	100%	99.5%	GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLS	95.8%	95.8%	100%	100%	99.8%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GNAL	100%	100%	100%	99.9%	99.3%	Dystonia 25, 615073

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

GNAO1	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	100%	100%	100%	100%	99.5%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GPAA1	88.5%	88.5%	100%	100%	99.4%	Glycosylphosphatidylinositol biosynthesis defect 15, 617810
GPR143	93.1%	93.1%	98.6%	89.3%	71.2%	Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814
GRIA4	100%	100%	100%	99.9%	99.5%	Neurodevelopmental disorder with or without seizures and gait abnormalities, 617864
GRID2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIK2	95.3%	95.3%	100%	99.9%	99.3%	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures, 619580;Intellectual developmental disorder, autosomal recessive 6, 611092

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

GRIN1	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2B	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRM1	100%	100%	100%	100%	99.6%	Spinocerebellar ataxia, autosomal recessive 13, 614831;Spinocerebellar ataxia 44, 617691
GRN	100%	100%	100%	100%	99.7%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
HACE1	95.9%	95.9%	100%	100%	99.7%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756

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

HEPACAM	100%	100%	100%	100%	99.5%	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925;Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without impaired intellectual development, 613926
HEXB	100%	100%	100%	99.9%	99.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HK1	98.2%	96.5%	100%	99.9%	99.5%	Anemia, congenital, nonspherocytic hemolytic, 5, hexokinase deficient, 235700;Retinitis pigmentosa 79, 617460;Neuropathy, hereditary motor and sensory, Russe type, 605285;Neurodevelopmental disorder with visual defects and brain anomalies, 618547
HNRNPR	100%	100%	100%	100%	99.9%	Neurodevelopmental disorder with dysmorphic facies and skeletal and brain abnormalities, 620073
HPCA	100%	100%	100%	99.9%	98.8%	Dystonia 2, torsion, autosomal recessive, 224500
HPDL	100%	100%	100%	100%	99.8%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026;Spastic paraplegia 83, autosomal recessive, 619027
HPRT1	87.6%	87.4%	99%	91.6%	74%	Hyperuricemia, HPRT-related, 300323;Lesch-Nyhan syndrome, 300322

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

HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSPD1	99.9%	98.9%	100%	100%	99.9%	Spastic paraplegia 13, autosomal dominant, 605280;Leukodystrophy, hypomyelinating, 4, 612233
HYCC1	100%	100%	100%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
IBA57	100%	100%	100%	100%	99.5%	Multiple mitochondrial dysfunctions syndrome 3, 615330;?Spastic paraplegia 74, autosomal recessive, 616451
IRF2BPL	100%	100%	99.8%	98.6%	94.6%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ISCA2	100%	100%	100%	100%	99.7%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITPR1	100%	100%	100%	100%	99.6%	Gillespie syndrome, 206700;Spinocerebellar ataxia 29, congenital nonprogressive, 117360;Spinocerebellar ataxia 15, 606658
JAM2	87.3%	87.3%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	98.1%	93.6%	100%	100%	99.5%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730

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

KARS1	94.3%	94.3%	100%	100%	99.8%	Deafness, autosomal recessive 89, 613916;Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147;?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641;Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196
KATNB1	100%	100%	100%	100%	99.6%	Lissencephaly 6, with microcephaly, 616212
KCNA1	100%	100%	100%	100%	99.5%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 32, 616366
KCNC1	100%	100%	100%	99.8%	99%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	100%	99.5%	99.8%	98.4%	93.4%	Spinocerebellar ataxia 13, 605259
KCND3	100%	100%	100%	99.9%	99%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNJ10	100%	100%	100%	100%	99.7%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNJ6	100%	100%	100%	100%	99.8%	Keppen-Lubinsky syndrome, 614098

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

KCNMA1	100%	100%	100%	99.9%	99.4%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729
KCNN2	100%	100%	100%	99.9%	98.9%	?Dystonia 34, myoclonic, 619724;Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725
KCTD17	100%	100%	100%	100%	99.1%	Dystonia 26, myoclonic, 616398
KCTD7	100%	100%	100%	100%	99.9%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIDINS220	100%	100%	100%	100%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF1A	100%	100%	100%	100%	99.3%	NESCAV syndrome, 614255;Neuropathy, hereditary sensory, type IIC, 614213;Spastic paraplegia 30, autosomal dominant, 610357;Spastic paraplegia 30, autosomal recessive, 620607
KIF1C	100%	100%	100%	100%	99.5%	Spastic ataxia 2, autosomal recessive, 611302

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

KIF5A	100%	100%	100%	100%	99.5%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
KMT2B	100%	99.7%	100%	99.9%	98.8%	Intellectual developmental disorder, autosomal dominant 68, 619934;Dystonia 28, childhood-onset, 617284
KPNA3	100%	100%	100%	100%	99.7%	Spastic paraplegia 88, autosomal dominant, 620106
L1CAM	100%	100%	98.4%	88.2%	69.2%	MASA syndrome, 303350;Hydrocephalus, congenital, X-linked, 307000;?Corpus callosum, partial agenesis of, 304100
LAMA1	98.9%	98.9%	100%	100%	99.6%	Poretti-Boltshauser syndrome, 615960
LAMB1	100%	100%	100%	100%	99.7%	Leukoencephalopathy without lacunae, adult-onset, 621424;Leukoencephalopathy with variable cortical brain malformations and/or hydrocephalus, 615191
LMNB1	100%	100%	100%	99.9%	99.2%	Microcephaly 26, primary, autosomal dominant, 619179
MAG	100%	100%	100%	100%	99.3%	Spastic paraplegia 75, autosomal recessive, 616680
MAPK8IP3	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443

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

MARS2	100%	100%	100%	99.9%	99.2%	?Combined oxidative phosphorylation deficiency 25, 616430;Spastic ataxia 3, autosomal recessive, 611390
MBIP	96.6%	96%	100%	100%	99.7%	
MECP2	100%	100%	99%	84.8%	65.9%	Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750
MECR	93.5%	93.5%	100%	100%	99.6%	?Optic atrophy 16, 620629;Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	89.1%	89.1%	100%	100%	99.9%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100%	100%	100%	100%	100%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MICU1	86.7%	86.7%	100%	100%	99.8%	Myopathy with extrapyramidal signs, 615673
MLC1	100%	100%	100%	100%	99.2%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004

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

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
MRE11	97.4%	97.4%	100%	100%	99.9%	Ataxia-telangiectasia-like disorder 1, 604391
MTHFR	100%	100%	100%	100%	99.5%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTHFS	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination, 618367
MTPAP	96.6%	96.6%	100%	100%	99.4%	?Spastic ataxia 4, autosomal recessive, 613672
MTRFR	100%	100%	100%	100%	99.7%	Spastic paraplegia 55, autosomal recessive, 615035;Combined oxidative phosphorylation deficiency 7, 613559
MTTP	100%	100%	100%	100%	99.8%	Abetalipoproteinemia, 200100
MYBPC1	100%	100%	100%	100%	99.8%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335
MYORG	100%	100%	100%	99.9%	99.1%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317

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

NANS	94.8%	94.3%	100%	99.9%	99.7%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NARS2	92.3%	92.3%	100%	100%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAXE	97.6%	93.5%	100%	100%	99.5%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NDUFS7	89.1%	85%	100%	100%	99.4%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NEFL	100%	100%	100%	99.9%	98.8%	Charcot-Marie-Tooth disease, type 1F, 607734;Charcot-Marie-Tooth disease, dominant intermediate G, 617882;Charcot-Marie-Tooth disease, type 2E, 607684
NEU1	100%	100%	100%	99.9%	99.5%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEXMIF	100%	100%	99.3%	91.3%	72.3%	Intellectual developmental disorder, X-linked 98, 300912
NF2	100%	100%	100%	100%	99.5%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000
NFU1	98.9%	94.6%	100%	100%	99.7%	Spastic paraplegia 93, autosomal recessive, 620938;Multiple mitochondrial dysfunctions syndrome 1, 605711

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

NGLY1	98.4%	97.2%	100%	100%	99.8%	Congenital disorder of deglycosylation 1, 615273
NIPA1	100%	100%	99.8%	99.2%	97.4%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	100%	100%	100%	99.9%	98.4%	Chorea, hereditary benign, 118700;{Thyroid cancer, nonmedullary, 1}, 188550;Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX6-2	100%	100%	100%	99.6%	97.1%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NPC1	100%	100%	100%	100%	99.7%	Niemann-Pick disease, type C1, 257220;Niemann-Pick disease, type D, 257220
NPC2	99%	95.2%	100%	100%	99.6%	Niemann-pick disease, type C2, 607625
NPTX1	100%	100%	100%	99.9%	98.5%	Spinocerebellar ataxia 50, 620158
NR4A2	100%	100%	100%	99.9%	99.4%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NT5C2	91.4%	91.4%	100%	100%	99.8%	Spastic paraplegia 45, autosomal recessive, 613162
NUBPL	89.8%	89.8%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 21, 618242

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

NUP62	100%	100%	100%	100%	99.3%	Striatonigral degeneration, infantile, 271930
NUS1	93.9%	93.9%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
OCLN	94.5%	94.5%	100%	99.9%	99.4%	Pseudo-TORCH syndrome 1, 251290
OGDHL	100%	100%	100%	100%	99.5%	
OPA1	94.7%	94.7%	100%	100%	99.7%	Optic atrophy plus syndrome, 125250;{Glaucoma, normal tension, susceptibility to}, 606657;Optic atrophy 1, 165500;Behr syndrome, 210000;Mitochondrial DNA depletion syndrome 14A (encephalomyopathic type), 621481;?Mitochondrial DNA depletion syndrome 14B (cardioencephalomyopathic type), 616896
OPHN1	94%	94%	98.9%	90.6%	72.9%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
PACS2	78.8%	78.8%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 66, 618067
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200

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

PAX6	100%	100%	100%	100%	99.5%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia /coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190
PCYT2	99.9%	97.1%	100%	100%	99.5%	Spastic paraplegia 82, autosomal recessive, 618770
PDE10A	100%	99.4%	99.9%	99.7%	98.3%	Striatal degeneration, autosomal dominant, 616922;Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE2A	100%	100%	100%	100%	99.3%	Intellectual developmental disorder with paroxysmal dyskinesia or seizures, 619150
PDE8B	100%	100%	100%	99.9%	99.4%	Pigmented nodular adrenocortical disease, primary, 3, 614190;Striatal degeneration, autosomal dominant, 609161
PDGFB	99.7%	94.7%	100%	100%	99.8%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483

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

PDGFRB	99.3%	97.4%	100%	100%	99.2%	Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PDHA1	91.4%	89.5%	99.3%	91.4%	72.9%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	91.6%	91.6%	100%	100%	99.6%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	100%	100%	100%	100%	99.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100%	100%	100%	100%	99.4%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	100%	100%	100%	100%	99.9%	Spinocerebellar ataxia 23, 610245
PEX10	100%	100%	100%	99.6%	98.7%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX2	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 5A (Zellweger), 614866;Peroxisome biogenesis disorder 5B, 614867
PEX7	97.9%	97.9%	100%	100%	99.6%	Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879

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

PHYH	100%	100%	100%	99.8%	99.1%	Refsum disease, 266500
PIGG	100%	100%	100%	100%	99.5%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLP1	94.5%	94.1%	98.8%	88.7%	69.7%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMP22	100%	100%	100%	100%	99.7%	Charcot-Marie-Tooth disease, type 1A, 118220;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, type 1E, 118300;?Neuropathy, inflammatory demyelinating, 139393;Neuropathy, recurrent, with pressure palsies, 162500;Dejerine-Sottas disease, 145900
PMPCA	96.2%	96.2%	100%	100%	99.1%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	100%	100%	100%	100%	99.4%	Paroxysmal nonkinesigenic dyskinesia 1, 118800

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

PNKP	100%	100%	100%	99.9%	99.2%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
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
POLG	100%	100%	100%	100%	99.8%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450

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

POLR1C	83.6%	83.5%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR3A	100%	100%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POU4F1	95.7%	92.7%	100%	99.5%	96.9%	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352
PPP1R3F	97%	97%	98.6%	88.8%	70.5%	
PPT1	90.3%	90.3%	100%	100%	99.7%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRDX3	100%	98.8%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 32, 619862;Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871
PRF1	100%	100%	100%	100%	99.6%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRICKLE1	100%	100%	100%	100%	99.8%	Epilepsy, progressive myoclonic 1B, 612437

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

PRKCG	100%	100%	100%	99.8%	98.3%	Spinocerebellar ataxia 14, 605361
PRKN	100%	100%	100%	100%	99.8%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	100%	99.8%	Dystonia 16, 612067
PRRT2	100%	100%	100%	100%	98.5%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200
PSAP	99.6%	97.1%	100%	100%	99.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PTRH2	100%	100%	100%	100%	99.9%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	100%	100%	100%	100%	99.6%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	92.4%	92.4%	100%	99.9%	99.2%	Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719

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

PYCR2	100%	100%	100%	100%	99.5%	Leukodystrophy, hypomyelinating, 10, 616420
QDPR	93.7%	93.6%	100%	100%	99.3%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	100%	100%	100%	100%	99.5%	Warburg micro syndrome 3, 614222
RAB3GAP1	95.7%	95.7%	100%	100%	99.6%	Martsolf syndrome 2, 619420;Warburg micro syndrome 1, 600118
RAB3GAP2	94.4%	94.4%	100%	100%	99.8%	Martsolf syndrome 1, 212720;Warburg micro syndrome 2, 614225
RAD51	89.4%	89.4%	100%	100%	99.5%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RARS1	90.9%	90.5%	100%	99.9%	99.4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	94.2%	93.8%	100%	100%	99.9%	Pontocerebellar hypoplasia, type 6, 611523
REEP1	87.5%	87.5%	100%	100%	99.7%	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011;Spastic paraplegia 31, autosomal dominant, 610250;?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751
REEP2	100%	100%	100%	99.8%	99.3%	Spastic paraplegia 72A, autosomal dominant, 615625;?Spastic paraplegia 72B, autosomal recessive, 620606

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

RFC1	100%	100%	100%	100%	99.7%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
RHOBTB2	98.7%	98.7%	100%	100%	99.6%	Developmental and epileptic encephalopathy 64, 618004
RNASEH2A	100%	100%	100%	100%	98.8%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.5%	91.5%	100%	99.9%	99.5%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	99.9%	98.6%	Aicardi-Goutieres syndrome 3, 610329
RNF170	100%	100%	100%	100%	99.9%	Ataxia, sensory, 1, autosomal dominant, 608984;Spastic paraplegia 85, autosomal recessive, 619686
RNF216	100%	100%	100%	100%	99.7%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RORA	100%	100%	100%	99.8%	98.6%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RTN2	100%	100%	100%	100%	99.2%	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854;Spastic paraplegia 12, autosomal dominant, 604805
RUBCN	100%	100%	100%	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	99%	99%	100%	100%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550

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

SAMD9L	100%	100%	100%	100%	99.9%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SAMHD1	92%	89.8%	100%	100%	99.5%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SCN11A	100%	100%	100%	99.8%	99.4%	Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317;Migraine, familial hemiplegic, 3, 609634;Dravet syndrome, 607208;Febrile seizures, familial, 3A, 604403;Generalized epilepsy with febrile seizures plus, type 2, 604403
SCN2A	100%	100%	100%	100%	99.8%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
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

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

SCYL1	100%	100%	100%	99.9%	99.1%	Spinocerebellar ataxia, autosomal recessive 21, 616719
SELENOI	100%	100%	100%	100%	99.9%	Spastic paraplegia 81, autosomal recessive, 618768
SEPSECS	99.4%	96.3%	100%	100%	99.5%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100%	100%	100%	100%	99.9%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	89.5%	89.5%	100%	100%	99.6%	Dystonia-11, myoclonic, 159900
SHQ1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with dystonia and seizures, 619922;?Dystonia 35, childhood-onset, 619921
SIL1	91.5%	91.5%	100%	100%	99.5%	Marinesco-Sjogren syndrome, 248800
SLC12A6	100%	100%	100%	100%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000;Charcot-Marie-Tooth disease, axonal, type 2II, 620068
SLC16A2	88.4%	88.4%	99%	91.3%	72%	Allan-Herndon-Dudley syndrome, 300523
SLC18A2	99.6%	96.7%	100%	100%	99.8%	Parkinsonism-dystonia, infantile, 2, 618049

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

SLC19A3	99.9%	98.9%	100%	100%	99.7%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A3	97.1%	94.3%	100%	100%	99.6%	Episodic ataxia, type 6, 612656
SLC1A4	100%	100%	100%	99.9%	99.3%	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657
SLC20A2	100%	99.5%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	100%	100%	100%	100%	99.8%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A46	100%	100%	100%	100%	99.7%	Neuropathy, hereditary motor and sensory, type VIB, 616505;Pontocerebellar hypoplasia, type 1E, 619303
SLC2A1	100%	100%	100%	100%	99.3%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC30A10	100%	100%	100%	100%	99.3%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	92.6%	92.6%	100%	100%	99.7%	Spastic paraplegia 42, autosomal dominant, 612539;Huppke-Brendel syndrome, 614482

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

SLC39A14	94%	94%	100%	100%	99.5%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC52A2	100%	100%	100%	99.9%	99.7%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	99.4%	?Fazio-Londe disease, 211500;Brown-Vialetto-Van Laere syndrome 1, 211530
SLC6A3	93.9%	92.4%	100%	100%	99.6%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SLC6A5	100%	100%	100%	100%	99.6%	Hyperekplexia 3, 614618
SLC9A1	100%	100%	100%	99.9%	99.1%	Lichtenstein-Knorr syndrome, 616291
SMDT1	100%	100%	100%	100%	99.9%	
SMPD1	100%	100%	100%	100%	99.6%	Niemann-Pick disease, type B, 607616;Niemann-Pick disease, type A, 257200
SNAPC4	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515
SNORD118						Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX14	95%	95%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 20, 616354

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

SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPART	95.8%	95.8%	100%	100%	99.7%	Troyer syndrome, 275900
SPAST	94.7%	94.6%	100%	100%	99.5%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100%	100%	100%	100%	99.7%	Mast syndrome, 248900
SPG7	100%	100%	100%	99.9%	99.3%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	100%	100%	100%	100%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716

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

SPTAN1	99.2%	98.8%	100%	100%	99.5%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528
SPTBN2	100%	100%	100%	99.9%	99.3%	Spinocerebellar ataxia 5, 600224;Spinocerebellar ataxia, autosomal recessive 14, 615386
SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261
STUB1	100%	100%	100%	100%	99.4%	Spinocerebellar ataxia 48, 618093;Spinocerebellar ataxia, autosomal recessive 16, 615768
SUCLA2	96.3%	96.3%	100%	100%	99.9%	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073

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

SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SUMF1	100%	100%	100%	100%	99.9%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	100%	99.5%	Sulfite oxidase deficiency, 272300
SYNE1	100%	100%	100%	100%	99.8%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743
TANGO2	100%	100%	100%	100%	99.6%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D20	92.2%	92.2%	100%	100%	99.7%	Warburg micro syndrome 4, 615663
TBC1D23	96.3%	96.3%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 11, 617695
TBCD	91.2%	90.7%	100%	99.9%	99.2%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TDP1	87.9%	87.5%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949

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

TECPR2	100%	100%	100%	99.9%	99.6%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TENM4	100%	100%	100%	100%	99.6%	Essential tremor, hereditary, 5, 616736
TH	100%	99.8%	100%	100%	99.2%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	100%	100%	99.8%	Dystonia 6, torsion, 602629
TIMM8A	100%	100%	98.2%	87.9%	71.6%	Mohr-Tranebjaerg syndrome, 304700
TMEM106B	100%	100%	100%	100%	99.9%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM240	100%	100%	100%	99.7%	97.6%	Spinocerebellar ataxia 21, 607454
TMEM67	96.1%	96.1%	100%	100%	99.7%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TOE1	100%	100%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 7, 614969
TOR1A	93.1%	90.7%	100%	99.9%	99.4%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TPP1	100%	100%	100%	100%	99.3%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270

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

TREM2	100%	100%	100%	100%	99.9%	{Alzheimer disease 17, susceptibility to}, 615080;Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRIT1	100%	100%	100%	100%	99.8%	Combined oxidative phosphorylation deficiency 35, 617873
TRPM3	97.8%	97.8%	100%	100%	99.6%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TSEN15	100%	100%	100%	100%	99.8%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100%	100%	99.6%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204

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

TSPOAP1	100%	100%	100%	100%	99.5%	Dystonia 22, juvenile-onset, 620453;?Dystonia 22, adult-onset, 620456
TTBK2	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia 11, 604432
TTC19	100%	100%	100%	100%	99.7%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	87.1%	87.1%	100%	100%	99.7%	Ataxia with isolated vitamin E deficiency, 277460
TUBA1A	100%	100%	100%	100%	99.6%	Lissencephaly 3, 611603
TUBA4A	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 23, 621231;Spastic ataxia 11, autosomal dominant, 621226;Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208;Congenital myopathy 26, 621225
TUBB	99.8%	98.9%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB4A	99.2%	96.6%	100%	100%	99%	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

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

TWNK	100%	100%	100%	100%	99.6%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TYROBP	91.4%	91.2%	100%	99.8%	99.2%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UBA5	94.1%	92.2%	100%	100%	99.8%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBAP1	96.8%	96.8%	100%	100%	99.5%	Spastic paraplegia 80, autosomal dominant, 618418
UBTF	100%	100%	100%	100%	99.4%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	100%	100%	100%	100%	99.8%	{?Parkinson disease 5, susceptibility to}, 613643;Spastic paraplegia 79A, autosomal dominant, 620221;Spastic paraplegia 79B, autosomal recessive, 615491
VAC14	96.4%	96.4%	100%	100%	99.7%	Striatonigral degeneration, childhood-onset, 617054
VAMP1	100%	100%	100%	99.9%	98.9%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600

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

VARS2	100%	100%	100%	100%	98.9%	Combined oxidative phosphorylation deficiency 20, 615917
VCP	100%	99.6%	100%	100%	99.8%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954;Charcot-Marie-Tooth disease, type 2Y, 616687;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VLDLR	100%	100%	100%	100%	99.8%	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050
VPS11	100%	100%	100%	100%	99.6%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100%	100%	100%	100%	99.9%	Choreoacanthocytosis, 200150
VPS13D	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100%	100%	100%	100%	99.6%	Dystonia 30, 619291
VPS37A	92.3%	92.3%	100%	99.9%	99.5%	Spastic paraplegia 53, autosomal recessive, 614898
VPS41	98.4%	96.8%	100%	100%	99.9%	Spinocerebellar ataxia, autosomal recessive 29, 619389
VPS53	83.1%	81.5%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99.6%	98%	100%	100%	99.9%	Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542

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

WASHC5	100%	100%	100%	100%	99.8%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563
WDR26	93.8%	93.8%	100%	100%	99.5%	Skraban-Deardorff syndrome, 617616
WDR45	98.4%	98%	98.4%	87.9%	68.6%	Neurodegeneration with brain iron accumulation 5, 300894
WDR73	100%	100%	100%	100%	99.3%	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
WFS1	91.5%	90.5%	100%	100%	99.4%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
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
XK	100%	99.9%	99.4%	92.2%	74%	McLeod syndrome, 300842

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

XPA	100%	100%	100%	100%	99.9%	Xeroderma pigmentosum, group A, 278700
XPR1	100%	100%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100%	100%	100%	100%	99.2%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
ZC4H2	100%	100%	98.3%	88.3%	72.7%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZFYVE26	100%	100%	100%	100%	99.7%	Spastic paraplegia 15, autosomal recessive, 270700

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