MOVEMENT DISORDERS GENE PANEL DG 3.4.0 (375 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100,0%	100,0%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCB7	99,7%	99,5%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	100,0%	100,0%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADAR	100,0%	100,0%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
ADCY5	100,0%	99,9%	Dyskinesia with orofacial involvement, autosomal dominant, 606703 Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 Dyskinesia with orofacial involvement, autosomal recessive, 619647
ADGRG1	100,0%	100,0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
ADPRS	100,0%	100,0%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	100,0%	100,0%	Spastic ataxia 5, autosomal recessive, 614487 Optic atrophy 12, 618977 Spinocerebellar ataxia 28, 610246
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
AGTPBP1	100,0%	100,0%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIMP1	100,0%	100,0%	Leukodystrophy, hypomyelinating, 3, 260600
ALDH18A1	100,0%	100,0%	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	93,2%	93,2%	Sjogren-Larsson syndrome, 270200
ALDH5A1	100,0%	100,0%	Succinic semialdehyde dehydrogenase deficiency, 271980

ALS2	100,0%	100,0%	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100
AMPD2	100,0%	100,0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
ANO10	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	100,0%	100,0%	Dystonia 24, 615034
AP4B1	100,0%	100,0%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100,0%	100,0%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	100,0%	100,0%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	87,9%	87,9%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	100,0%	100,0%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	92,9%	92,9%	Argininemia, 207800
ARSA	100,0%	100,0%	Metachromatic leukodystrophy, 250100
ARX	99,0%	96,8%	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	100,0%	100,0%	Canavan disease, 271900
ATCAY	100,0%	100,0%	Ataxia, cerebellar, Cayman type, 601238
ATL1	100,0%	100,0%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATM	100,0%	100,0%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, T-cell prolymphocytic leukemia, somatic, Lymphoma, mantle cell, somatic,
ATP13A2	100,0%	100,0%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A2	100,0%	100,0%	Developmental and epileptic encephalopathy 98, 619605 Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235

			CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
ATP2B3	100,0%	100,0%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	100,0%	100,0%	Wilson disease, 277900
B4GALNT1	100,0%	100,0%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	100,0%	100,0%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	100,0%	100,0%	Maple syrup urine disease, type Ia, 248600
BCKDHB	100,0%	100,0%	Maple syrup urine disease, type Ib, 248600
BCL11B	100,0%	99,9%	Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 Immunodeficiency 49, 617237
BRAT1	100,0%	100,0%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BSCL2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	83,1%	83,1%	Biotinidase deficiency, 253260
C19orf12	100,0%	100,0%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CA8	100,0%	100,0%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	100,0%	100,0%	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	100,0%	100,0%	Developmental and epileptic encephalopathy 69, 618285
CACNA1G	100,0%	100,0%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNB4	100,0%	100,0%	Episodic ataxia, type 5, 613855
CAMTA1	100,0%	100,0%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CAPN1	100,0%	100,0%	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	100,0%	100,0%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHMP1A	100,0%	100,0%	Pontocerebellar hypoplasia, type 8, 614961
CLCN2	100,0%	100,0%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN4	100,0%	100,0%	Raynaud-Claes syndrome, 300114
CLN5	71,7%	71,6%	Ceroid lipofuscinosis, neuronal, 5, 256731

CLN6	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 Ceroid lipofuscinosis, neuronal, 6A, 601780
CLP1	100,0%	100,0%	Pontocerebellar hypoplasia, type 10, 615803
CLPB	100,0%	100,0%	Neutropenia, severe congenital, 9, autosomal dominant, 619813 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
COASY	100,0%	100,0%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
COL4A1	100,0%	100,0%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564 Brain small vessel disease with or without ocular anomalies, 175780
COL4A2	100,0%	100,0%	Brain small vessel disease 2, 614483
COL6A1	100,0%	100,0%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	100,0%	100,0%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100,0%	100,0%	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COQ2	97,2%	97,2%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	100,0%	100,0%	Mitochondrial complex IV deficiency, nuclear type 11, 619054
CP	100,0%	100,0%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CSF1R	100,0%	100,0%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
CSTB	100,0%	100,0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTBP1	100,0%	99,4%	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915
CTSD	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CWF19L1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 17, 616127
CYP27A1	100,0%	100,0%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	100,0%	100,0%	Spastic paraplegia 56, autosomal recessive, 615030

CYP7B1	100,0%	100,0%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DARS1	100,0%	100,0%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100,0%	100,0%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	100,0%	100,0%	Maple syrup urine disease, type II, 248600
DCAF17	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080
DCC	100,0%	100,0%	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	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605
DDC	100,0%	100,0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	100,0%	100,0%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100,0%	100,0%	Spastic paraplegia 54, autosomal recessive, 615033
DHDDS	95,2%	95,2%	Developmental delay and seizures with or without movement abnormalities, 617836 ?Congenital disorder of glycosylation, type 1bb, 613861 Retinitis pigmentosa 59, 613861
DLAT	100,0%	100,0%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100,0%	100,0%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAJC12	100,0%	100,0%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384
DNAJC3	100,0%	100,0%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAL4	100,0%	100,0%	?Mirror movements 3, 616059
DNM1L	100,0%	100,0%	Optic atrophy 5, 610708 Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DNMT1	100,0%	99,7%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DPYS	100,0%	100,0%	Dihydropyrimidinuria, 222748
DTYMK	100,0%	100,0%	No OMIM Disease ID
EBF3	100,0%	100,0%	Hypotonia, ataxia, and delayed development syndrome, 617330
ECHS1	100,0%	100,0%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2B1	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	100,0%	100,0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896

EIF2B5	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
ELOVL4	100,0%	100,0%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	100,0%	100,0%	Spinocerebellar ataxia 38, 615957
ERLIN2	100,0%	100,0%	Spastic paraplegia 18, autosomal recessive, 611225
ETHE1	100,0%	100,0%	Ethylmalonic encephalopathy, 602473
EXOSC3	100,0%	100,0%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100,0%	100,0%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
EXOSC8	100,0%	100,0%	Pontocerebellar hypoplasia, type 1C, 616081
EXOSC9	100,0%	100,0%	Pontocerebellar hypoplasia, type 1D, 618065
FA2H	100,0%	100,0%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	100,0%	100,0%	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	100,0%	100,0%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 Cataracts, spastic paraparesis, and speech delay, 619338
FARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FBXO7	100,0%	100,0%	Parkinson disease 15, autosomal recessive, 260300
FGF14	100,0%	100,0%	Spinocerebellar ataxia 27, 609307
FLVCR1	100,0%	100,0%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	100,0%	100,0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	100,0%	99,8%	Nystagmus, infantile periodic alternating, X-linked, 310700 Nystagmus 1, congenital, X-linked, 310700
FTL	100,0%	100,0%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GALC	100,0%	100,0%	Krabbe disease, 245200
GAMT	100,0%	100,0%	Cerebral creatine deficiency syndrome 2, 612736
GAN	100,0%	100,0%	Giant axonal neuropathy-1, 256850
GBA	100,0%	100,0%	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
GBA2	100,0%	100,0%	Spastic paraplegia 46, autosomal recessive, 614409

GBE1	100,0%	100,0%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GCDH	100,0%	100,0%	Glutaricaciduria, type I, 231670
GCH1	100,0%	100,0%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GDAP2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GFAP	100,0%	100,0%	Alexander disease, 203450
GJC2	99,9%	99,5%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GLB1	100,0%	100,0%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GNAL	100,0%	100,0%	Dystonia 25, 615073
GOSR2	100,0%	100,0%	Epilepsy, progressive myoclonic 6, 614018
GPR143	100,0%	100,0%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GRID2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIN1	100,0%	100,0%	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	100,0%	100,0%	Developmental and epileptic encephalopathy 27, 616139 Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRM1	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 13, 614831 Spinocerebellar ataxia 44, 617691
GRN	100,0%	100,0%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
HACE1	100,0%	100,0%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HEXB	100,0%	100,0%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HK1	100,0%	100,0%	Retinitis pigmentosa 79, 617460 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Hemolytic anemia due to hexokinase deficiency, 235700
HPCA	100,0%	100,0%	Dystonia 2, torsion, autosomal recessive, 224500
HPDL	100,0%	100,0%	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 Spastic paraplegia 83, autosomal recessive, 619027

HPRT1	100,0%	100,0%	Hyperuricemia, HRPT-related, 300323 Lesch-Nyhan syndrome, 300322
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPD1	100,0%	100,0%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IBA57	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, autosomal recessive, 616451
IRF2BPL	100,0%	100,0%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ISCA2	100,0%	100,0%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITPR1	100,0%	100,0%	Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360 Spinocerebellar ataxia 15, 606658
JAM2	92,3%	92,3%	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824
JAM3	100,0%	100,0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KATNB1	100,0%	100,0%	Lissencephaly 6, with microcephaly, 616212
KCNA1	100,0%	100,0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100,0%	100,0%	Developmental and epileptic encephalopathy 32, 616366
KCNC1	100,0%	100,0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	99,8%	98,8%	Spinocerebellar ataxia 13, 605259
KCND3	100,0%	100,0%	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399
KCNJ10	100,0%	100,0%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	100,0%	100,0%	Keppen-Lubinsky syndrome, 614098
KCNMA1	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 Cerebellar atrophy, developmental delay, and seizures, 617643 Liang-Wang syndrome, 618729
KCTD7	100,0%	100,0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIDINS220	100,0%	100,0%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 Ventriculomegaly and arthrogryposis, 619501
KIF1A	98,0%	98,0%	NESCAV syndrome, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal dominant, 610357 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	100,0%	100,0%	Spastic ataxia 2, autosomal recessive, 611302

KIF5A	100,0%	100,0%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KMT2B	99,7%	99,3%	Dystonia 28, childhood-onset, 617284
L1CAM	100,0%	100,0%	MASA syndrome, 303350 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus due to aqueductal stenosis, 307000
LAMA1	100,0%	100,0%	Poretti-Boltshauser syndrome, 615960
LAMB1	100,0%	100,0%	Lissencephaly 5, 615191
LMNB1	100,0%	100,0%	Leukodystrophy, adult-onset, autosomal dominant, 169500 Microcephaly 26, primary, autosomal dominant, 619179
MAG	100,0%	100,0%	Spastic paraplegia 75, autosomal recessive, 616680
MAPK8IP3	100,0%	100,0%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MARS2	100,0%	100,0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MECP2	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Encephalopathy, neonatal severe, 300673 Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MECR	100,0%	100,0%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	100,0%	100,0%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100,0%	100,0%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MICU1	100,0%	100,0%	Myopathy with extrapyramidal signs, 615673
MLC1	100,0%	100,0%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMADHC	89,7%	89,7%	Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Homocystinuria, cblD type, variant 1, 277410
MRE11	100,0%	100,0%	Ataxia-telangiectasia-like disorder 1, 604391
MTHFR	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MTPAP	100,0%	100,0%	?Spastic ataxia 4, autosomal recessive, 613672
C12orf65	100,0%	100,0%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
MTTP	100,0%	100,0%	Abetalipoproteinemia, 200100

MYORG	100,0%	100,0%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
NANS	100,0%	100,0%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 24, 616239 ?Deafness, autosomal recessive 94, 618434
NDUFS7	100,0%	100,0%	Mitochondrial complex I deficiency, nuclear type 3, 618224
NEFL	100,0%	100,0%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684
NEU1	100,0%	100,0%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100,0%	100,0%	Intellectual developmental disorder, X-linked 98, 300912
NF2	100,0%	100,0%	Neurofibromatosis, type 2, 101000 Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091
NGLY1	100,0%	100,0%	Congenital disorder of deglycosylation 1, 615273
NIPA1	100,0%	100,0%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	100,0%	100,0%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX6-2	100,0%	100,0%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NOL3	100,0%	100,0%	?Myoclonus, familial, 1, 614937
NPC1	100,0%	100,0%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	100,0%	100,0%	Niemann-pick disease, type C2, 607625
NT5C2	100,0%	100,0%	Spastic paraplegia 45, autosomal recessive, 613162
NUP62	100,0%	100,0%	Striatonigral degeneration, infantile, 271930
OCLN	100,0%	100,0%	Pseudo-TORCH syndrome 1, 251290
OPA1	100,0%	100,0%	Optic atrophy plus syndrome, 125250 Optic atrophy 1, 165500 Behr syndrome, 210000 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPHN1	100,0%	99,5%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
PACS2	100,0%	100,0%	Developmental and epileptic encephalopathy 66, 618067
PANK2	100,0%	100,0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX6	100,0%	100,0%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200

			?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PCYT2	100,0%	100,0%	Spastic paraplegia 82, autosomal recessive, 618770
PDE10A	90,0%	87,2%	Striatal degeneration, autosomal dominant, 616922 Dyskinesia, limb and orofacial, infantile-onset, 616921
PDE8B	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	100,0%	100,0%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRB	100,0%	100,0%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007
PDHA1	100,0%	100,0%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	100,0%	100,0%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	97,4%	97,4%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	100,0%	100,0%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	100,0%	100,0%	Spinocerebellar ataxia 23, 610245
PEX10	100,0%	100,0%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX2	100,0%	100,0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	100,0%	100,0%	Refsum disease, 266500
PIK3R5	100,0%	100,0%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
PLP1	100,0%	100,0%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	100,0%	100,0%	Congenital disorder of glycosylation, type Ia, 212065

PMP22	100,0%	100,0%	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	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	100,0%	100,0%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100,0%	100,0%	?Charcot-Marie-Tooth disease, type 2B2, 605589 Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPLA6	100,0%	100,0%	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POLG	100,0%	100,0%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR1C	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR3A	100,0%	100,0%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
PPT1	82,5%	82,5%	Ceroid lipofuscinosis, neuronal, 1, 256730
PRF1	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PRICKLE1	100,0%	100,0%	Epilepsy, progressive myoclonic 1B, 612437
PRKCG	100,0%	100,0%	Spinocerebellar ataxia 14, 605361
PRKRA	100,0%	100,0%	Dystonia 16, 612067
PRRT2	100,0%	100,0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Seizures, benign familial infantile, 2, 605751 Episodic kinesigenic dyskinesia 1, 128200
PSAP	100,0%	100,0%	Combined SAP deficiency, 611721 Krabbe disease, atypical, 611722

			Metachromatic leukodystrophy due to SAP-b deficiency, 249900 Gaucher disease, atypical, 610539
PTRH2	100,0%	100,0%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	100,0%	100,0%	Spinocerebellar ataxia 47, 617931
PYCR2	100,0%	100,0%	Leukodystrophy, hypomyelinating, 10, 616420
QDPR	100,0%	100,0%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	100,0%	100,0%	Warburg micro syndrome 3, 614222
RAB3GAP1	99,4%	99,4%	Martsolf syndrome 2, 619420 Warburg micro syndrome 1, 600118
RAB3GAP2	100,0%	100,0%	Martsolf syndrome 1, 212720 Warburg micro syndrome 2, 614225
RAD51	89,4%	89,4%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RARS1	94,4%	94,4%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100,0%	100,0%	Pontocerebellar hypoplasia, type 6, 611523
REEP1	100,0%	100,0%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RNASEH2A	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
RNF170	100,0%	100,0%	Ataxia, sensory, 1, autosomal dominant, 608984 Spastic paraplegia 85, autosomal recessive, 619686
RNF216	100,0%	100,0%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	100,0%	100,0%	Spastic paraplegia 12, autosomal dominant, 604805
RUBCN	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	100,0%	100,0%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100,0%	100,0%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCN11A	100,0%	100,0%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN1A	100,0%	100,0%	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	100,0%	100,0%	Seizures, benign familial infantile, 3, 607745 Developmental and epileptic encephalopathy 11, 613721 Episodic ataxia, type 9, 618924
SCN8A	100,0%	100,0%	?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 Developmental and epileptic encephalopathy 13, 614558
SEPSECS	100,0%	100,0%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	100,0%	100,0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	91,2%	91,2%	Dystonia-11, myoclonic, 159900
SIL1	100,0%	100,0%	Marinesco-Sjogren syndrome, 248800
SLC12A6	100,0%	100,0%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	100,0%	100,0%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	98,7%	98,7%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	100,0%	100,0%	Episodic ataxia, type 6, 612656
SLC20A2	100,0%	100,0%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	100,0%	100,0%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC2A1	100,0%	100,0%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC30A10	100,0%	100,0%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	100,0%	100,0%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC39A14	93,6%	93,5%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC52A2	100,0%	100,0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100,0%	100,0%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC6A3	100,0%	100,0%	Parkinsonism-dystonia, infantile, 1, 613135
SLC9A1	100,0%	100,0%	Lichtenstein-Knorr syndrome, 616291
SMDT1	100,0%	100,0%	No OMIM Disease ID

SMPD1	100,0%	100,0%	Niemann-Pick disease, type B, 607616 Niemann-Pick disease, type A, 257200
SNCA	79,1%	79,1%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNORD118	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX14	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SPART	100,0%	100,0%	Troyer syndrome, 275900
SPAST	100,0%	100,0%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	100,0%	100,0%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	100,0%	100,0%	Mast syndrome, 248900
SPG7	100,0%	100,0%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	100,0%	100,0%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	100,0%	99,9%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	100,0%	100,0%	Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
SUMF1	100,0%	100,0%	Multiple sulfatase deficiency, 272200
SUOX	100,0%	100,0%	Sulfite oxidase deficiency, 272300
SYNE1	98,8%	98,8%	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TAF1	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TANGO2	100,0%	100,0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D20	100,0%	100,0%	Warburg micro syndrome 4, 615663
TBC1D23	100,0%	100,0%	Pontocerebellar hypoplasia, type 11, 617695
TBCD	100,0%	100,0%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TDP1	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100,0%	100,0%	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031
TENM4	100,0%	100,0%	Essential tremor, hereditary, 5, 616736

TGM6	100,0%	100,0%	Spinocerebellar ataxia 35, 613908
TH	100,0%	100,0%	Segawa syndrome, recessive, 605407
THAP1	100,0%	100,0%	Dystonia 6, torsion, 602629
TIMM8A	100,0%	100,0%	Mohr-Tranebjaerg syndrome, 304700
TMEM106B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM240	100,0%	100,0%	Spinocerebellar ataxia 21, 607454
TMEM67	100,0%	100,0%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TOE1	100,0%	100,0%	Pontocerebellar hypoplasia, type 7, 614969
TOR1A	92,9%	91,5%	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TPP1	100,0%	100,0%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREM2	100,0%	100,0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRPM3	100,0%	100,0%	No OMIM Disease ID
TSEN15	100,0%	100,0%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100,0%	100,0%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100,0%	100,0%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TTBK2	100,0%	100,0%	Spinocerebellar ataxia 11, 604432
TTC19	100,0%	100,0%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	100,0%	100,0%	Ataxia with isolated vitamin E deficiency, 277460
TUBA1A	100,0%	100,0%	Lissencephaly 3, 611603
TUBB	100,0%	99,8%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TUBB4A	99,5%	97,4%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100,0%	100,0%	Cortical dysplasia, complex, with other brain malformations 4, 615412

TWNK	100,0%	100,0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYROBP	100,0%	100,0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UBAP1	100,0%	100,0%	Spastic paraplegia 80, autosomal dominant, 618418
UBTF	100,0%	100,0%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UCHL1	100,0%	100,0%	Spastic paraplegia 79, autosomal recessive, 615491
VAMP1	100,0%	100,0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VARS2	100,0%	100,0%	Combined oxidative phosphorylation deficiency 20, 615917
VCP	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VLDLR	100,0%	100,0%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS11	100,0%	100,0%	?Dystonia 32, 619637 Leukodystrophy, hypomyelinating, 12, 616683
VPS13A	100,0%	100,0%	Choreoacanthocytosis, 200150
VPS13D	100,0%	100,0%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100,0%	100,0%	Dystonia 30, 619291
VPS37A	100,0%	100,0%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	100,0%	99,8%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	100,0%	100,0%	Pontocerebellar hypoplasia type 1A, 607596
WASHC5	100,0%	100,0%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WDR26	97,0%	94,3%	Skraban-Deardorff syndrome, 617616
WDR45	100,0%	100,0%	Neurodegeneration with brain iron accumulation 5, 300894
WDR73	100,0%	100,0%	Galloway-Mowat syndrome 1, 251300
WDR81	100,0%	100,0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WFS1	100,0%	100,0%	Deafness, autosomal dominant 6/14/38, 600965 ?Cataract 41, 116400 Wolfram-like syndrome, autosomal dominant, 614296 Wolfram syndrome 1, 222300
WWOX	100,0%	100,0%	Esophageal squamous cell carcinoma, somatic, 133239 Developmental and epileptic encephalopathy 28, 616211 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	100,0%	100,0%	McLeod syndrome with or without chronic granulomatous disease, 300842

XPR1	100,0%	100,0%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	100,0%	100,0%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
ZC4H2	100,0%	100,0%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZFYVE26	100,0%	100,0%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100,0%	100,0%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	100,0%	100,0%	No OMIM Disease ID

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.
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 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors