Bevegelsesforstyrrelser

Genpanel, versjon v01

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

Gen	Transkript	>10x	Fenotype
ABCB7	NM_004299.4	100%	Anemia, sideroblastic, with ataxia OMIM
ABCD1	NM_000033.3	76%	Adrenoleukodystrophy OMIM Adrenomyeloneuropathy, adult OMIM
ABHD12	NM_001042472.2	99%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract OMIM
ACAT1	NM_000019.3	100%	Alpha-methylacetoacetic aciduria OMIM
ADCK3	NM_020247.4	100%	Coenzyme Q10 deficiency, primary, 4 OMIM
ADCY5	NM_183357.2	98%	Dyskinesia, familial, with facial myokymia OMIM
AFG3L2	NM_006796.2	98%	Ataxia, spastic, 5, autosomal recessive OMIM Spinocerebellar ataxia 28 OMIM
ALDH18A1	NM_002860.3	100%	Cutis laxa, autosomal dominant 3 OMIM Cutis laxa, autosomal recessive, type IIIA OMIM Spastic paraplegia 9A, autosomal dominant OMIM Spastic paraplegia 9B, autosomal recessive OMIM
ALDH3A2	NM_000382.2	100%	Sjogren-Larsson syndrome OMIM
ALS2	NM_020919.3	100%	Amyotrophic lateral sclerosis 2, juvenile OMIM Primary lateral sclerosis, juvenile OMIM Spastic paralysis, infantile onset ascending OMIM
AMN	NM_030943.3	98%	Adrenoleukodystrophy OMIM Adrenomyeloneuropathy, adult OMIM Megaloblastic anemia-1, Norwegian type OMIM
AMPD2	NM_001257360.1	100%	?Spastic paraplegia 63 OMIM Pontocerebellar hypoplasia, type 9 OMIM
AMT	NM_000481.3	100%	Glycine encephalopathy OMIM
ANO10	NM_018075.3	100%	Spinocerebellar ataxia, autosomal recessive 10 OMIM

Gen	Transkript	>10x	Fenotype
ANO3	NM_031418.2	100%	Dystonia 24 OMIM
AP4B1	NM_006594.3	100%	Spastic paraplegia 47, autosomal recessive OMIM
AP4E1	NM_007347.4	100%	Spastic paraplegia 51, autosomal recessive OMIM
AP4M1	NM_004722.3	99%	Spastic paraplegia 50, autosomal recessive OMIM
AP4S1	NM_007077.4	100%	Spastic paraplegia 52, autosomal recessive OMIM
AP5Z1	NM_014855.2	100%	Spastic paraplegia 48, autosomal recessive OMIM
APTX	NM_175073.2	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia OMIM
ARG1	NM_000045.3	100%	Argininemia OMIM
ARSA	NM_000487.5	100%	Metachromatic leukodystrophy OMIM
ATCAY	NM_033064.4	100%	Ataxia, cerebellar, Cayman type OMIM
ATL1	NM_015915.4	100%	Neuropathy, hereditary sensory, type ID OMIM Spastic paraplegia 3A, autosomal dominant OMIM
ATM	NM_000051.3	100%	Ataxia-telangiectasia OMIM
ATP13A2	NM_022089.3	100%	?Ceroid lipofuscinosis, neuronal, 12 OMIM Kufor-Rakeb syndrome OMIM
ATP1A2	NM_000702.3	100%	Alternating hemiplegia of childhood OMIM Migraine, familial basilar OMIM Migraine, familial hemiplegic, 2 OMIM
ATP1A3	NM_152296.4	100%	Alternating hemiplegia of childhood 2 OMIM CAPOS syndrome OMIM Dystonia-12 OMIM
ATP2B3	NM_001001344.2	99%	?Spinocerebellar ataxia, X-linked 1 OMIM
ATP7B	NM_000053.3	100%	Wilson disease OMIM
AUH	NM_001698.2	100%	3-methylglutaconic aciduria, type I OMIM

Gen	Transkript	>10x	Fenotype
B4GALNT1	NM_001478.4	100%	Spastic paraplegia 26, autosomal recessive OMIM
BCAP31	NM_001139441.1	98%	Deafness, dystonia, and cerebral hypomyelination OMIM
BCKDHA	NM_000709.3	100%	Maple syrup urine disease, type Ia OMIM
BCKDHB	NM_183050.2	99%	Maple syrup urine disease, type Ib OMIM
BICD2	NM_001003800.1	100%	Spinal muscular atrophy, lower extremity-predominant, 2, AD OMIM
BSCL2	NM_032667.6	100%	Encephalopathy, progressive, with or without lipodystrophy OMIM Lipodystrophy, congenital generalized, type 2 OMIM Neuropathy, distal hereditary motor, type VA OMIM Silver spastic paraplegia syndrome OMIM
C10orf2	NM_021830.4	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) OMIM Perrault syndrome 5 OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3 OMIM
C12orf65	NM_152269.4	100%	Combined oxidative phosphorylation deficiency 7 OMIM Spastic paraplegia 55, autosomal recessive OMIM
C19orf12	NM_001031726.3	100%	?Spastic paraplegia 43, autosomal recessive OMIM Neurodegeneration with brain iron accumulation 4 OMIM
CA8	NM_004056.4	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 OMIM
CACNA1A	NM_001127221.1	99%	Epileptic encephalopathy, early infantile, 42 OMIM Episodic ataxia, type 2 OMIM Migraine, familial hemiplegic, 1 OMIM Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia OMIM Spinocerebellar ataxia 6 OMIM
CACNA1G	NM_018896.4	100%	Spinocerebellar ataxia 42 OMIM
CACNB4	NM_000726.3	100%	Episodic ataxia, type 5 OMIM
CAPN1	NM_001198868.1	100%	Spastic paraplegia 76, autosomal recessive OMIM
CASK	NM_003688.3	100%	FG syndrome 4 OMIM Mental retardation and microcephaly with pontine and cerebellar hypoplasia OMIM Mental retardation, with or without nystagmus OMIM
CBS	NM_000071.2	100%	Homocystinuria, B6-responsive and nonresponsive types OMIM Thrombosis, hyperhomocysteinemic OMIM

Gen	Transkript	>10x	Fenotype
CCT5	NM_012073.3	100%	Neuropathy, hereditary sensory, with spastic paraplegia OMIM
CHMP1A	NM_002768.4	100%	Pontocerebellar hypoplasia, type 8 OMIM
CLP1	NM_006831.2	100%	Pontocerebellar hypoplasia, type 10 OMIM
COASY	NM_025233.6	100%	Neurodegeneration with brain iron accumulation 6 OMIM
COQ2	NM_015697.7	100%	Coenzyme Q10 deficiency, primary, 1 OMIM
COQ9	NM_020312.3	100%	Coenzyme Q10 deficiency, primary, 5 OMIM
CP	NM_000096.3	97%	Cerebellar ataxia OMIM Hemosiderosis, systemic, due to aceruloplasminemia OMIM
CSF1R	NM_005211.3	99%	Leukoencephalopathy, diffuse hereditary, with spheroids OMIM
CUBN	NM_001081.3	100%	Megaloblastic anemia-1, Finnish type OMIM
CYP27A1	NM_000784.3	100%	Cerebrotendinous xanthomatosis OMIM
CYP2U1	NM_183075.2	99%	Spastic paraplegia 56, autosomal recessive OMIM
CYP7B1	NM_004820.3	100%	Spastic paraplegia 5A, autosomal recessive OMIM
DARS2	NM_018122.4	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation OMIM
DBT	NM_001918.3	100%	Maple syrup urine disease, type II OMIM
DCAF17	NM_025000.3	100%	Woodhouse-Sakati syndrome OMIM
DCTN1	NM_004082.4	100%	Neuropathy, distal hereditary motor, type VIIB OMIM Perry syndrome OMIM
DDC	NM_000790.3	99%	Aromatic L-amino acid decarboxylase deficiency OMIM
DDHD1	NM_001160147.1	99%	Spastic paraplegia 28, autosomal recessive OMIM

Gen	Transkript	>10x	Fenotype
DDHD2	NM_015214.2	100%	Spastic paraplegia 54, autosomal recessive OMIM
DLAT	NM_001931.4	100%	Pyruvate dehydrogenase E2 deficiency OMIM
DNMT1	NM_001130823.1	99%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant OMIM Neuropathy, hereditary sensory, type IE OMIM
EIF2B1	NM_001414.3	100%	Leukoencephalopathy with vanishing white matter OMIM
EIF2B2	NM_014239.3	100%	Leukoencephalopathy with vanishing white matter OMIM Ovarioleukodystrophy OMIM
EIF2B3	NM_020365.4	100%	Leukoencephalopathy with vanishing white matter OMIM
EIF2B4	NM_015636.3	100%	Leukoencephaly with vanishing white matter OMIM Ovarioleukodystrophy OMIM
EIF2B5	NM_003907.2	100%	Leukoencephalopathy with vanishing white matter OMIM Ovarioleukodystrophy OMIM
ELOVL5	NM_021814.4	100%	Spinocerebellar ataxia 38 OMIM
ENTPD1	NM_001776.5	100%	Spastic paraplegia 64, autosomal recessive OMIM
ERLIN2	NM_007175.6	100%	Spastic paraplegia 18, autosomal recessive OMIM
EXOSC3	NM_016042.3	99%	Pontocerebellar hypoplasia, type 1B OMIM
EXOSC8	NM_181503.2	100%	Pontocerebellar hypoplasia, type 1C OMIM
FA2H	NM_024306.4	100%	Spastic paraplegia 35, autosomal recessive OMIM
FAR1	NM_032228.5	99%	Peroxisomal fatty acyl-CoA reductase 1 disorder OMIM
FBXO7	NM_012179.3	100%	Parkinson disease 15, autosomal recessive OMIM
FGF14	NM_004115.3	100%	Spinocerebellar ataxia 27 OMIM
FLVCR1	NM_014053.3	100%	Ataxia, posterior column, with retinitis pigmentosa OMIM

Gen	Transkript	>10x	Fenotype
FOLR1	NM_016725.2	100%	Neurodegeneration due to cerebral folate transport deficiency OMIM
FTL	NM_000146.3	100%	Hyperferritinemia-cataract syndrome OMIM Neurodegeneration with brain iron accumulation 3 OMIM
FXN	NM_000144.4	99%	Friedreich ataxia OMIM Friedreich ataxia with retained reflexes OMIM
GALC	NM_000153.3	100%	Krabbe disease OMIM
GAMT	NM_000156.5	100%	Cerebral creatine deficiency syndrome 2 OMIM
GAN	NM_022041.3	100%	Giant axonal neuropathy-1 OMIM
GBA	NM_001005741.2	100%	Gaucher disease, perinatal lethal OMIM Gaucher disease, type I OMIM Gaucher disease, type II OMIM Gaucher disease, type III OMIM Gaucher disease, type IIIC OMIM
GBA2	NM_020944.2	100%	Spastic paraplegia 46, autosomal recessive OMIM
GCDH	NM_000159.3	100%	Glutaricaciduria, type I OMIM
GCH1	NM_000161.2	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia OMIM Hyperphenylalaninemia, BH4-deficient, B OMIM
GCSH	NM_004483.4	95%	Glycine encephalopathy OMIM
GFAP	NM_002055.4	100%	Alexander disease OMIM
GIF	NM_005142.2	100%	Intrinsic factor deficiency OMIM
GJC2	NM_020435.3	99%	Leukodystrophy, hypomyelinating, 2 OMIM Spastic paraplegia 44, autosomal recessive OMIM
GLB1	NM_000404.2	100%	GM1-gangliosidosis, type I OMIM GM1-gangliosidosis, type II OMIM GM1-gangliosidosis, type III OMIM Mucopolysaccharidosis type IVB (Morquio) OMIM
GLDC	NM_000170.2	97%	Glycine encephalopathy OMIM
GNAL	NM_001142339.2	100%	Dystonia 25 OMIM

Gen	Transkript	>10x	Fenotype
GOSR2	NM_004287.3	100%	Epilepsy, progressive myoclonic 6 OMIM
GRID2	NM_001510.3	100%	Spinocerebellar ataxia, autosomal recessive 18 OMIM
GRM1	NM_001278066.1	100%	Spinocerebellar ataxia, autosomal recessive 13 OMIM
HEXB	NM_000521.3	100%	Sandhoff disease, infantile, juvenile, and adult forms OMIM
HMGCL	NM_000191.2	100%	HMG-CoA lyase deficiency OMIM
HMGCS2	NM_005518.3	100%	HMG-CoA synthase-2 deficiency OMIM
HPRT1	NM_000194.2	99%	Lesch-Nyhan syndrome OMIM
HSD17B10	NM_004493.2	100%	?Mental retardation, X-linked syndromic 10 OMIM 17-beta-hydroxysteroid dehydrogenase X deficiency OMIM
HSPD1	NM_002156.4	99%	Leukodystrophy, hypomyelinating, 4 OMIM Spastic paraplegia 13, autosomal dominant OMIM
ITPR1	NM_002222.5	100%	Gillespie syndrome OMIM Spinocerebellar ataxia 15 OMIM Spinocerebellar ataxia 29, congenital nonprogressive OMIM
KCNA1	NM_000217.2	100%	Episodic ataxia/myokymia syndrome OMIM
KCNC3	NM_004977.2	97%	Spinocerebellar ataxia 13 OMIM
KCND3	NM_004980.4	100%	Brugada syndrome 9 OMIM Spinocerebellar ataxia 19 OMIM
KCNJ10	NM_002241.4	100%	Enlarged vestibular aqueduct, digenic OMIM SESAME syndrome OMIM
KCNMA1	NM_002247.3	100%	Generalized epilepsy and paroxysmal dyskinesia OMIM
KIAA0196	NM_014846.3	100%	Ritscher-Schinzel syndrome 1 OMIM Spastic paraplegia 8, autosomal dominant OMIM
KIF1A	NM_004321.6	100%	Mental retardation, autosomal dominant 9 OMIM Neuropathy, hereditary sensory, type IIC OMIM Spastic paraplegia 30, autosomal recessive OMIM
KIF1C	NM_006612.5	100%	Spastic ataxia 2, autosomal recessive OMIM

Gen	Transkript	>10x	Fenotype
KIF5A	NM_004984.2	100%	Spastic paraplegia 10, autosomal dominant OMIM
L1CAM	NM_000425.4	99%	Corpus callosum, partial agenesis of OMIM CRASH syndrome OMIM Hydrocephalus due to aqueductal stenosis OMIM Hydrocephalus with congenital idiopathic intestinal pseudoobstruction OMIM Hydrocephalus with Hirschsprung disease OMIM MASA syndrome OMIM
LMBRD1	NM_018368.3	99%	Methylmalonic aciduria and homocystinuria, cblF type OMIM
MAG	NM_002361.3	100%	Spastic paraplegia 75, autosomal recessive OMIM
MICU1	NM_006077.3	100%	Myopathy with extrapyramidal signs OMIM
MMAA	NM_172250.2	100%	Methylmalonic aciduria, vitamin B12-responsive OMIM
MMAB	NM_052845.3	100%	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type OMIM
MMACHC	NM_015506.2	100%	Methylmalonic aciduria and homocystinuria, cblC type OMIM
MRE11A	NM_005591.3	100%	Ataxia-telangiectasia-like disorder OMIM
MTHFR	NM_005957.4	100%	Homocystinuria due to MTHFR deficiency OMIM
MTPAP	NM_018109.3	100%	Ataxia, spastic, 4 OMIM
MTR	NM_000254.2	100%	Homocystinuria-megaloblastic anemia, cblG complementation type OMIM
MTRR	NM_002454.2	100%	{Neural tube defects, folate-sensitive, susceptibility to} OMIM Homocystinuria-megaloblastic anemia, cbl E type OMIM
MTTP	NM_000253.3	100%	Abetalipoproteinemia OMIM
MUT	NM_000255.3	100%	Methylmalonic aciduria, mut(0) type OMIM
NIPA1	NM_144599.4	100%	Spastic paraplegia 6, autosomal dominant OMIM
NKX2-1	NM_001079668.2	100%	Chorea, hereditary benign OMIM

Gen	Transkript	>10x	Fenotype
NOL3	NM_001276312.1	99%	Myoclonus, familial cortical OMIM
NPC1	NM_000271.4	100%	Niemann-Pick disease, type C1 OMIM Niemann-Pick disease, type D OMIM
NPC2	NM_006432.3	100%	Niemann-pick disease, type C2 OMIM
NT5C2	NM_012229.4	100%	Spastic paraplegia 45, autosomal recessive OMIM
NUP62	NM_001193357.1	100%	Striatonigral degeneration, infantile OMIM
OPA1	NM_015560.2	100%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) OMIM Behr syndrome OMIM Optic atrophy 1 OMIM Optic atrophy plus syndrome OMIM
OPHN1	NM_002547.2	100%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance OMIM
OXCT1	NM_000436.3	100%	Succinyl CoA:3-oxoacid CoA transferase deficiency OMIM
PANK2	NM_153638.2	100%	HARP syndrome OMIM Neurodegeneration with brain iron accumulation 1 OMIM
PARK2	NM_004562.2	100%	Parkinson disease, juvenile, type 2 OMIM
PARK7	NM_007262.4	100%	Parkinson disease 7, autosomal recessive early-onset OMIM
PCBD1	NM_000281.3	100%	Hyperphenylalaninemia, BH4-deficient, D OMIM
PCCA	NM_000282.3	99%	Propionicacidemia OMIM
PCCB	NM_000532.4	100%	Propionicacidemia OMIM
PDE10A	NM_001130690.2	100%	Dyskinesia, limb and orofacial, infantile-onset OMIM Striatal degeneration, autosomal dominant OMIM
PDE8B	NM_003719.3	100%	Pigmented nodular adrenocortical disease, primary, 3 OMIM Striatal degeneration, autosomal dominant OMIM
PDGFB	NM_002608.2	100%	Basal ganglia calcification, idiopathic, 5 OMIM
PDGFRB	NM_002609.3	99%	Basal ganglia calcification, idiopathic, 4 OMIM

Gen	Transkript	>10x	Fenotype
PDHA1	NM_000284.3	99%	Pyruvate dehydrogenase E1-alpha deficiency OMIM
PDHX	NM_003477.2	100%	Lacticacidemia due to PDX1 deficiency OMIM
PDSS1	NM_014317.3	99%	Coenzyme Q10 deficiency, primary, 2 OMIM
PDSS2	NM_020381.3	99%	Coenzyme Q10 deficiency, primary, 3 OMIM
PDYN	NM_024411.4	100%	Spinocerebellar ataxia 23 OMIM
PEX10	NM_153818.1	100%	Peroxisome biogenesis disorder 6A (Zellweger) OMIM Peroxisome biogenesis disorder 6B OMIM
PGAP1	NM_024989.3	100%	Mental retardation, autosomal recessive 42 OMIM
PHYH	NM_006214.3	100%	Refsum disease OMIM
PIK3R5	NM_001142633.2	100%	Ataxia-oculomotor apraxia 3 OMIM
PLA2G6	NM_003560.2	100%	Infantile neuroaxonal dystrophy 1 OMIM Neurodegeneration with brain iron accumulation 2B OMIM Parkinson disease 14, autosomal recessive OMIM
PLP1	NM_000533.4	100%	Pelizaeus-Merzbacher disease OMIM Spastic paraplegia 2, X-linked OMIM
PNKD	NM_015488.4	100%	Paroxysmal nonkinesigenic dyskinesia OMIM
PNPLA6	NM_006702.4	99%	?Laurence-Moon syndrome OMIM Boucher-Neuhauser syndrome OMIM Oliver-McFarlane syndrome OMIM Spastic paraplegia 39, autosomal recessive OMIM
POLG	NM_002693.2	100%	Mitochondrial DNA depletion syndrome 4A (Alpers type) OMIM Mitochondrial DNA depletion syndrome 4B (MNGIE type) OMIM Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE) OMIM Progressive external ophthalmoplegia, autosomal dominant 1 OMIM Progressive external ophthalmoplegia, autosomal recessive 1 OMIM
POLR3A	NM_007055.3	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
POLR3B	NM_018082.5	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism OMIM
PRKCG	NM_002739.3	100%	Spinocerebellar ataxia 14 OMIM

Gen	Transkript	>10x	Fenotype
PRKRA	NM_003690.4	100%	Dystonia 16 OMIM
PRRT2	NM_145239.2	100%	Convulsions, familial infantile, with paroxysmal choreoathetosis OMIM Episodic kinesigenic dyskinesia 1 OMIM Seizures, benign familial infantile, 2 OMIM
PTS	NM_000317.2	100%	Hyperphenylalaninemia, BH4-deficient, A OMIM
QDPR	NM_000320.2	100%	Hyperphenylalaninemia, BH4-deficient, C OMIM
RARS2	NM_020320.3	100%	Pontocerebellar hypoplasia, type 6 OMIM
REEP1	NM_022912.2	100%	?Neuronopathy, distal hereditary motor, type VB OMIM Spastic paraplegia 31, autosomal dominant OMIM
RNF170	NM_001160223.1	100%	Ataxia, sensory, 1, autosomal dominant OMIM
RTN2	NM_005619.4	100%	Spastic paraplegia 12, autosomal dominant OMIM
SACS	NM_014363.5	100%	Spastic ataxia, Charlevoix-Saguenay type OMIM
SAMHD1	NM_015474.3	100%	Aicardi-Goutieres syndrome 5 OMIM
SEPSECS	NM_016955.3	100%	Pontocerebellar hypoplasia type 2D OMIM
SERAC1	NM_032861.3	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome OMIM
SETX	NM_015046.5	100%	Amyotrophic lateral sclerosis 4, juvenile OMIM Spinocerebellar ataxia, autosomal recessive 1 OMIM
SGCE	NM_003919.2	100%	Dystonia-11, myoclonic OMIM
SIL1	NM_022464.4	100%	Marinesco-Sjogren syndrome OMIM
SLC16A2	NM_006517.4	98%	Allan-Herndon-Dudley syndrome OMIM
SLC19A3	NM_025243.3	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) OMIM
SLC1A3	NM_004172.4	100%	Episodic ataxia, type 6 OMIM

Gen	Transkript	>10x	Fenotype
SLC20A2	NM_006749.4	100%	Basal ganglia calcification, idiopathic, 1 OMIM
SLC25A15	NM_014252.3	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome OMIM
SLC2A1	NM_006516.2	100%	Dystonia 9 OMIM GLUT1 deficiency syndrome 1, infantile onset, severe OMIM GLUT1 deficiency syndrome 2, childhood onset OMIM Stomatin-deficient cryohydrocytosis with neurologic defects OMIM
SLC30A10	NM_018713.2	100%	Hypermanganesemia with dystonia 1 OMIM
SLC33A1	NM_004733.3	100%	Congenital cataracts, hearing loss, and neurodegeneration OMIM Spastic paraplegia 42, autosomal dominant OMIM
SLC52A2	NM_024531.4	100%	Brown-Vialetto-Van Laere syndrome 2 OMIM
SLC52A3	NM_033409.3	100%	Brown-Vialetto-Van Laere syndrome 1 OMIM Fazio-Londe disease OMIM
SLC6A3	NM_001044.4	100%	Parkinsonism-dystonia, infantile OMIM
SLC9A6	NM_006359.2	100%	Mental retardation, X-linked syndromic, Christianson type OMIM
SMPD1	NM_000543.4	100%	Niemann-Pick disease, type A OMIM Niemann-Pick disease, type B OMIM
SNCA	NM_000345.3	100%	Dementia, Lewy body OMIM Parkinson disease 1 OMIM Parkinson disease 4 OMIM
SNX14	NM_020468.5	100%	Spinocerebellar ataxia, autosomal recessive 20 OMIM
SPAST	NM_014946.3	100%	Spastic paraplegia 4, autosomal dominant OMIM
SPG11	NM_025137.3	100%	Amyotrophic lateral sclerosis 5, juvenile OMIM Charcot-Marie-Tooth disease, axonal, type 2X OMIM Spastic paraplegia 11, autosomal recessive OMIM
SPG20	NM_015087.4	100%	Troyer syndrome OMIM
SPG21	NM_016630.6	100%	Mast syndrome OMIM
SPG7	NM_003119.3	99%	Spastic paraplegia 7, autosomal recessive OMIM
SPR	NM_003124.4	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency OMIM

Gen	Transkript	>10x	Fenotype
SPTBN2	NM_006946.2	100%	Spinocerebellar ataxia 5 OMIM Spinocerebellar ataxia, autosomal recessive 14 OMIM
STUB1	NM_005861.3	100%	Spinocerebellar ataxia, autosomal recessive 16 OMIM
SUOX	NM_000456.2	100%	Sulfite oxidase deficiency OMIM
SYNE1	NM_033071.3	100%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant OMIM Spinocerebellar ataxia, autosomal recessive 8 OMIM
TAF1	NM_004606.4	99%	Dystonia-Parkinsonism, X-linked OMIM Mental retardation, X-linked, syndromic 33 OMIM
TDP1	NM_018319.3	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy OMIM
TECPR2	NM_014844.3	100%	Spastic paraplegia 49, autosomal recessive OMIM
TGM6	NM_198994.2	99%	Spinocerebellar ataxia 35 OMIM
TH	NM_199292.2	99%	Segawa syndrome, recessive OMIM
THAP1	NM_018105.2	100%	Dystonia 6, torsion OMIM
TIMM8A	NM_004085.3	96%	Mohr-Tranebjaerg syndrome OMIM
TMEM240	NM_001114748.1	100%	Spinocerebellar ataxia 21 OMIM
TMEM67	NM_153704.5	99%	COACH syndrome OMIM Joubert syndrome 6 OMIM Meckel syndrome 3 OMIM Nephronophthisis 11 OMIM
TOR1A	NM_000113.2	100%	Dystonia-1, torsion OMIM
TPP1	NM_000391.3	100%	Ceroid lipofuscinosis, neuronal, 2 OMIM Spinocerebellar ataxia, autosomal recessive 7 OMIM
TSEN2	NM_025265.3	100%	Pontocerebellar hypoplasia type 2B OMIM
TSEN54	NM_207346.2	99%	?Pontocerebellar hypoplasia type 5 OMIM Pontocerebellar hypoplasia type 2A OMIM Pontocerebellar hypoplasia type 4 OMIM
TTBK2	NM_173500.3	100%	Spinocerebellar ataxia 11 OMIM

Gen	Transkript	>10x	Fenotype
TTC19	NM_017775.3	98%	Mitochondrial complex III deficiency, nuclear type 2 OMIM
TTPA	NM_000370.3	100%	Ataxia with isolated vitamin E deficiency OMIM
TUBB4A	NM_006087.3	100%	Dystonia 4, torsion, autosomal dominant OMIM Leukodystrophy, hypomyelinating, 6 OMIM
VAMP1	NM_014231.4	100%	Spastic ataxia 1, autosomal dominant OMIM
VLDLR	NM_003383.3	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 OMIM
VPS13A	NM_033305.2	99%	Choreoacanthocytosis OMIM
VPS37A	NM_152415.2	99%	Spastic paraplegia 53, autosomal recessive OMIM
VPS53	NM_001128159.2	100%	Pontocerebellar hypoplasia, type 2E OMIM
VRK1	NM_003384.2	100%	Pontocerebellar hypoplasia type 1A OMIM
WDR45	NM_007075.3	99%	Neurodegeneration with brain iron acculation 5 OMIM
WDR81	NM_001163809.1	100%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 OMIM
WWOX	NM_016373.3	100%	Epileptic encephalopathy, early infantile, 28 OMIM Esophageal squamous cell carcinoma, somatic OMIM Spinocrebellar ataxia, autosomal recessive 12 OMIM
XK	NM_021083.2	100%	McLeod syndrome with or without chronic granulomatous disease OMIM
XPR1	NM_004736.3	100%	Basal ganglia calcification, idiopathic, 6 OMIM
ZFYVE26	NM_015346.3	100%	Spastic paraplegia 15, autosomal recessive OMIM