Bewegingsstoornisssen genpanel v1 (203 genen)

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
ABCB7	99,8	Anemia, sideroblastic, with ataxia, 301310
ABCD1	98,7	Adrenoleukodystrophy, 300100
		Adrenomyeloneuropathy, adult, 300100
ABHD12	93,8	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	100,0	Dystonia, juvenile-onset, 607371
		Baraitser-Winter syndrome 1, 243310
ADCK3	99,7	Coenzyme Q10 deficiency, primary, 4, 612016
ADCY5	99,6	Dyskinesia, familial, with facial myokymia, 606703
ADGRG1	100,0	Polymicrogyria, bilateral frontoparieta, 606854
		Polymicrogyria, bilateral perisylvian, 615752
AFG3L2	95,0	Spinocerebellar ataxia 28, 610246
		Ataxia, spastic, 5, autosomal recessive, 614487
ALDH3A2	100,0	Sjogren-Larsson syndrome, 270200
ANO10	99,8	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	99,7	Dystonia 24, 615034
AP4B1	99,4	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	100,0	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,9	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	96,3	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	100,0	Spastic paraplegia 48, autosomal recessive, 613647
APTX	60,5	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARSA	100,0	Metachromatic leukodystrophy, 250100
ARX	90,2	Epileptic encephalopathy, early infantile, 1, 308350
		Lissencephaly, X-linked 2, 300215
		Mental retardation, X-linked 29 and others, 300419
		Proud syndrome, 300004
		Partington syndrome, 309510
		Hydranencephaly with abnormal genitalia, 300215
ASPA	100,0	Canavan disease, 271900
ATCAY	100,0	Ataxia, cerebellar, Cayman type, 601238
ATL1	100,0	Spastic paraplegia 3A, autosomal dominant, 182600
		Neuropathy, hereditary sensory, type ID, 613708
ATM	100,0	Ataxia-telangiectasia, 208900
		Lymphoma, B-cell non-Hodgkin, somatic
		{Breast cancer, susceptibility to}, 114480
		Lymphoma, mantle cell
		T-cell prolymphocytic leukemia, somati
ATP13A2	99,0	Parkinson disease 9, 606693
ATP1A3	100,0	Dystonia-12, 128235
		Alternating hemiplegia of childhood 2, 614820
ATP2B3	99,5	Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	100,0	Wilson disease, 277900
B4GALNT1	100,0	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	94,0	Deafness, dystonia and cerebellar hypomyelination, 300475
BCKDHA	100,0	Maple syrup urine disease, type Ia, 248600
BCKDHB	100,0	Maple syrup urine disease, type Ib, 248600
BSCL2	77,5	Lipodystrophy, congenital generalized, type 2, 269700
		Silver spastic paraplegia syndrome, 270685
		Neuropathy, distal hereditary motor, type V, 600794
C10orf2	100,0	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245
		Progressive external ophthalmoplegia, autosomal dominant, 3, 609286
C12orf65	100,0	Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	100,0	Neurodegeneration with brain iron accumulation 4, 614298
CA8	100,0	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
CACNA1A	100,0	Migraine, familial hemiplegic, 1, 141500
		Episodic ataxia, type 2, 108500
		Spinocerebellar ataxia 6, 183086
		Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNB4	99,9	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
		{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682
		Episodic ataxia, type 5, 613855
CCT5	100,0	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	99,4	Dystonia 23, 614860
COASY	98,5	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	100,0	Coenzyme Q10 deficiency, primary, 1, 607426
		{Multiple system atrophy, susceptibility to}, 146500
COQ9	97,3	Coenzyme Q10 deficiency, primary, 5, 614654
CP	93,5	[Hypoceruloplasminemia, hereditary], 604290
		Cerebellar ataxia, 604290
		Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CSTB	100,0	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	97,7	Cerebrotendinous xanthomatosis, 213700
CYP2U1	94,5	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	99,8	Bile acid synthesis defect, congenital, 3, 613812
		Spastic paraplegia 5A, autosomal recessive, 270800
DBT	98,7	Maple syrup urine disease, type II, 248600
DCAF17	35,1	Woodhouse-Sakati syndrome, 241080
DCTN1	90,9	Neuropathy, distal hereditary motor, type VIIB, 607641
		{Amyotrophic lateral sclerosis, susceptibility to}, 105400
		Perry syndrome, 168605
DDC	100,0	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	99,9	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100,0	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	100,0	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	100,0	Dihydrolipoamide dehydrogenase deficiency, 246900
DNMT1	98,2	Neuropathy, hereditary sensory, type IE, 614116
		Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
EIF2B1	100,0	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	100,0	Leukoencephalopathy with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EIF2B3	99,3	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100,0	Leukoencephaly with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EIF2B5	100,0	Leukoencephalopathy with vanishing white matter, 603896
		Ovarioleukodystrophy, 603896
EIF4G1	100,0	Parkinson disease 18, 614251
ELOVL5	100,0	Spinocerebellar ataxia 38, 615957
ERLIN2	100,0	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	100,0	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	100,0	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBXO7	100,0	Parkinson disease 15, autosomal recessive, 260300
FGF14	99,3	Spinocerebellar ataxia 27, 609307
FLVCR1	99,4	Ataxia, posterior column, with retinitis pigmentosa, 609033
FTL	100,0	Hyperferritinemia-cataract syndrome, 600886
		Neurodegeneration with brain iron accumulation 3, 606159
GALC	99,9	Krabbe disease, 245200
GAN	99,7	Giant axonal neuropathy-1, 256850
GBA	99,7	Gaucher disease, type I, 230800
		Gaucher disease, type II, 230900
		Gaucher disease, type III, 231000
		Gaucher disease, type IIIC, 231005
		Gaucher disease, perinatal lethal, 608013

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
GBA2	100,0	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	87,5	Glutaricaciduria, type I, 231670
GCH1	100,0	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
		Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	99,6	Alexander disease, 203450
GJC2	92,5	Leukodystrophy, hypomyelinating, 2, 608804
		Spastic paraplegia 44, autosomal recessive, 613206
		Lymphedema, hereditary, IC, 613480
GLB1	100,0	GM1-gangliosidosis, type I, 230500
		GM1-gangliosidosis, type II, 230600
		GM1-gangliosidosis, type III, 230650
		Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	92,4	Dystonia 25, 615073
GOSR2	98,4	Epilepsy, progressive myoclonic 6, 614018
GRID2	100,0	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRM1	99,9	Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	100,0	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	93,4	Lesch-Nyhan syndrome, 300322
		HPRT-related gout, 300323
HSPD1	100,0	Spastic paraplegia 13, autosomal dominant, 605280
		Leukodystrophy, hypomyelinating, 4, 612233
ITPR1	100,0	Spinocerebellar ataxia 15, 606658
		Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	100,0	Episodic ataxia/myokymia syndrome, 160120
KCNC1	98,4	Epilepsy, progressive myoclonic 7, 616187
KCNC3	37,6	Spinocerebellar ataxia 13, 605259
KCNJ10	100,0	SESAME syndrome, 612780
		Enlarged vestibular aqueduct, digenic, 600791
KCTD7	99,9	Epilepsy,progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	100,0	Spastic paraplegia 8, autosomal dominant, 603563
KIF1A	100,0	Spastic paraplegia 30, autosomal recessive, 610357
		Neuropathy, hereditary sensory, type IIC, 614213
		Mental retardation, autosomal dominant 9, 614255
KIF1C	99,6	Spastic ataxia 2,autosomal recessive, 611302
KIF5A	100,0	Spastic paraplegia 10, autosomal dominant, 604187
L1CAM	99,8	Hydrocephalus due to aqueductal stenosis, 307000
		MASA syndrome, 303350
		CRASH syndrome, 303350
		Hydrocephalus with Hirschsprung disease, 307000
		Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000
		Corpus callosum, partial
MARS2	100,0	Spastic ataxia 3, autosomal recessive, 611390
MECP2	100,0	Rett syndrome, 312750
		Mental retardation, X-linked, syndromic 13, 300055
		Rett syndrome, preserved speech variant, 312750
		Encephalopathy, neonatal severe, 300673
		{Autism susceptibility, X-linked 3}, 300496
		Angelman syndrome, 105830
MICU1	99,6	Myopathy with extrapyramidal signs
MMADHC	100,0	Homocystinuria, cblD type, variant 1, 277410
		Methylmalonic aciduria, cblD type, variant 2, 277410
		Methylmalonic aciduria and homocystinuria, cblD type, 277410
MRE11A	99,9	Ataxia-telangiectasia-like disorder, 604391
MTHFR	99,9	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500
		{Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634
MTPAP	100,0	Ataxia, spastic, 4, 613672
MTTP	99,9	Abetalipoproteinemia, 200100
		{Metabolic syndrome, protection against}, 605552

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
NIPA1	99,3	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	100,0	Goiter, familial, due to TTF-1 defect (1)
		Chorea, hereditary benign, 118700
		Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NOL3	100,0	Myoclonus, familial cortical, 614937
NPC1	100,0	Niemann-Pick disease, type C1, 257220
		Niemann-Pick disease, type D, 257220
NPC2	100,0	Niemann-pick disease, type C2, 607625
NUP62	100,0	Striatonigral degeneration, infantile, 271930
OPA1	100,0	Optic atrophy 1, 165500
		{Glaucoma, normal tension, susceptibility to}, 606657
		Optic atrophy plus syndrome, 125250
PANK2	100,0	Neurodegeneration with brain iron accumulation 1, 234200
		HARP syndrome, 607236
PAX6	100,0	Aniridia, 106210
		Peters anomaly, 604229
		Cataract with late-onset corneal dystrophy, 106210
		Keratitis, 148190
		Foveal hyperplasia, 136520
		Morning glory disc anomaly, 120430
		Optic nerve hypoplasia, 165550
		Coloboma, ocular, 120200
		Coloboma of
PDE8B	99,9	Pigmented nodular adrenocortical disease, primary, 3, 614190
		Striatal degeneration, autosomal dominant, 609161
PDGFB	99,7	Basal ganglia calcification, idiopathic, 5, 615483
PDGFRB	99,6	Basal ganglia calcification, idiopathic, 4, 615007
		Myeloproliferative disorder with eosinophilia, 131440
		Myofibromatosis, infantile, 1, 228550
PDHA1	99,9	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	100,0	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	89,6	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,9	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	99,9	Spinocerebellar ataxia 23, 610245
PEX10	98,8	Peroxisome biogenesis disorder 6A (Zellweger), 614870
		Peroxisome biogenesis disorder 6B, 614871
PEX7	99,8	Rhizomelic chondrodysplasia punctata, type 1, 215100
		Peroxisome biogenesis disorder 9B, 614879
PHYH	100,0	Refsum disease, 266500
PIK3R5	100,0	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	100,0	Infantile neuroaxonal dystrophy 1, 256600
		Neurodegeneration with brain iron accumulation 2B, 610217
		Parkinson disease 14, 612953
PLP1	99,7	Pelizaeus-Merzbacher disease, 312080
		Spastic paraplegia 2, X-linked, 312920
PMM2	100,0	Congenital disorder of glycosylation, type Ia, 212065
PNKD	99,2	Paroxysmal nonkinesigenic dyskinesia, 118800
PNKP	100,0	Ataxia-oculomotor apraxia 4, 616267
		Microcephaly, seizures and developmental delay, 613402
PNPLA6	100,0	Spastic paraplegia 39, autosomal recessive, 612020
POLG	100,0	Progressive external ophthalmoplegia, autosomal recessive, 258450
		Progressive external ophthalmoplegia, autosomal dominant, 157640
		Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662
		Mitochondrial DNA depletion syndrome 4A (Alpers type), 20
PRKCG	99,8	Spinocerebellar ataxia 14, 605361
PRKRA	99,8	Dystonia 16, 612067
PRRT2	99,7	Episodic kinesigenic dyskinesia 1, 128200
		Seizures, benign familial infantile, 2, 605751

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
		Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
REEP1	100,0	Spastic paraplegia 31, autosomal dominant, 610250
		Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	100,0	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,0	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0	Aicardi-Goutieres syndrome 3, 610329
RNF170	28,5	taxia, sensory, 1, autosomal dominant, 608984
RTN2	100,0	Spastic paraplegia 12, autosomal dominant, 604805
RUBCN/ KIAA0226	98,4	?Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	100,0	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	99,9	Aicardi-Goutieres syndrome 5, 612952
		Chilblain lupus 2, 614415 -3
SCN8A	100,0	Cognitive impairment with or without cerebellar ataxia, 614306
		Epileptic encephalopathy, early infantile, 13, 614558
SERAC1	89,4	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	100,0	Ataxia-ocular apraxia-2, 606002
		Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	98,6	maternally imprinted Dystonia-11, myoclonic, 159900
SIL1	99,0	Marinesco-Sjogren syndrome, 248800
SLC12A6	99,9	Agenesis of coprus callosum with peripheral neuropathy, 218000
SLC16A2	100,0	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	99,9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	100,0	Episodic ataxia, type 6, 612656
SLC20A2	100,0	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	99,9	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC2A1	97,4	GLUT1 deficiency syndrome 1, 606777
		GLUT1 deficiency syndrome 2, 612126
		{Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847
		Dystonia 9, 601042
SLC30A10	57,9	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	100,0	Spastic paraplegia 42, autosomal dominant, 612539
		Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC52A2	68,9	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC6A3	100,0	{Nicotine dependence, protection against}, 188890
		Parkinsonism-dystonia, infantile, 613135
SLC9A1	43,8	No OMIM disease phenotype
SMPD1	99,9	Niemann-Pick disease, type A, 257200
		Niemann-Pick disease, type B, 607616
SNCA	100,0	Parkinson disease 4, 605543
		Dementia, Lewy body, 127750
		Parkinson disease 1, 168601
SNX14	99,2	Spinocerebellar ataxia, autosomal recessive 20, 616354
SPAST	99,9	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	99,9	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	100,0	Troyer syndrome, 275900
SPG21	100,0	Mast syndrome, 248900
SPG7	99,7	Spastic paraplegia 7, autosomal recessive, 607259
SPR	100,0	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	99,3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	100,0	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	99,6	Sulfite oxidase deficiency, 272300
SYNE1	99,9	Spinocerebellar ataxia, autosomal recessive 8, 610743
		Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TAF1	79,9	Dystonia-Parkinsonism, X-linked, 314250
TDP1	100,0	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	99,8	Spastic paraplegia 49, autosomal recessive, 615031

Gene	% covered > 30x	Associated Phenotype description and OMIM ID
TGM6	100,0	Spinocerebellar ataxia 35, 613908
TH	100,0	Segawa syndrome, recessive, 605407
THAP1	98,3	Dystonia 6, torsion, 602629
TIMM8A	99,9	Deafness, X-linked 1, progressive
		Mohr-Tranebjaerg syndrome, 304700
		Jensen syndrome, 311150
TMEM240	93,9	Spinocerebellar ataxia 21, 607454
TMEM67	93,6	Meckel syndrome 3, 607361
		Joubert syndrome 6, 610688
		{Bardet-Biedl syndrome 14, modifier of}, 209900
		COACH syndrome, 216360
		Nephronophthisis 11, 613550
TOR1A	100,0	Dystonia-1, torsion, 128100
		Dystonia, early-onset atypical, with myoclonic features
		{Dystonia-1, modifier of}
TREX1	100,0	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750
		Chilblain lupus, 610448
		Vasculopathy, retinal, with cerebral leukodystrophy, 192315
		{Systemic lupus erythematosus, susceptibility to}, 152700
TTBK2	100,0	Spinocerebellar ataxia 11, 604432
TTC19	96,9	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	99,9	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	100,0	?Dystonia 4, torsion, autosomal dominant, 128101
		Leukodystrophy, hypomyelinating, 6, 612438
VAMP1	58,4	spastic ataxia 1, autosomal dominant
VCP	98,1	Spastic Ataxia 1, autosomal dominant, 108600
VLDLR	97,4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	100,0	Choreoacanthocytosis, 200150
VPS37A	99,9	Spastic paraplegia 53, autosomal recessive, 614898
WDR45	100,0	?Neurodegeneration with brain iron accululation 5, 300894
WDR81	100,0	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	79,6	Esophageal squamous cell carcinoma, 133239
		Spinocerebellar ataxia 12, 614322
XPR1	94,3	Basal ganglia calcification, idiopathic, 6, 616413
ZFYVE26	99,9	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	99,8	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	100,0	Spinocerebellar ataxia, autosomal recessive 5, 606937

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Genes in bold are core genes
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
"No OMIM phenotype" signifies a gene without a current OMIM association
OMIM phenotype descriptions between {} signify risk factors