MOVEMENT DISORDERS GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCB7	61.3	100%	98%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	38.8	73%	68%	Adrenoleukodystrophy, 300100; Adrenomyeloneuropathy, adult, 300100
ABHD12	63.6	100%	97%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	66.9	100%	97%	Dystonia, juvenile-onset, 607371; Baraitser-Winter syndrome 1, 243310
ADCK3	110.2	100%	94%	Coenzyme Q10 deficiency, primary, 4, 612016
AFG3L2	83.5	100%	94%	Spinocerebellar ataxia 28, 610246; Ataxia, spastic, 5, autosomal recessive, 614487
ALDH3A2	104.2	100%	100%	Sjogren-Larsson syndrome, 270200
ANO10	112.0	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	116.4	100%	100%	Dystonia 24, 615034
AP4B1	111.4	100%	98%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	133.9	100%	99%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	106.8	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	77.8	100%	100%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	85.5	99%	89%	Spastic paraplegia 48, autosomal recessive, 613647
APTX	134.4	100%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARSA	94.6	97%	95%	Metachromatic leukodystrophy, 250100
ARX	33.7	76%	61%	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	116.3	100%	100%	Canavan disease, 271900
ATCAY	104.9	100%	100%	Ataxia, cerebellar, Cayman type, 601238
ATL1	114.3	100%	99%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATM	125.3	100%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somati

ATP13A2	84.8	99%	94%	Parkinson disease 9, 606693
ATP1A3	119.0	100%	99%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2B3	62.1	98%	92%	Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	133.8	99%	97%	Wilson disease, 277900
B4GALNT1	86.9	97%	89%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	36.8	71%	62%	Deafness, dystonia and cerebellar hypomyelination, 300475
BCKDHA	110.0	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	94.9	97%	94%	Maple syrup urine disease, type Ib, 248600
BSCL2	116.8	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
C10orf2	153.6	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia, autosomal dominant, 3, 609286
C12orf65	199.7	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035
C19ORF12	94.0	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298
CA8	90.4	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	83.0	98%	90%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNB4	99.8	100%	96%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CCT5	82.2	93%	86%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	113.1	98%	94%	Dystonia 23, 614860
COQ2	78.7	98%	87%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ9	94.6	96%	89%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	90.2	98%	94%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CSTB	186.4	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	123.5	100%	95%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	126.6	100%	97%	Spastic paraplegia 56, autosomal recessive, 615030

CYP7B1	105.0	100%	98%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	118.0	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	104.0	99%	95%	Woodhouse-Sakati syndrome, 241080
DCTN1	125.0	100%	97%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DDC	90.0	99%	98%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	148.0	99%	98%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	119.8	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	106.0	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	148.4	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNMT1	109.1	97%	95%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
EIF2B1	104.2	100%	98%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	106.2	100%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	95.8	100%	98%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	130.1	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	106.3	100%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	116.9	100%	99%	Parkinson disease 18, 614251
ELOVL5	86.8	100%	99%	Spinocerebellar ataxia 38, 615957
ERLIN2	124.5	99%	96%	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	63.8	94%	87%	Spastic paraplegia 35, autosomal recessive, 612319
FBXO7	157.9	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FGF14	121.1	100%	100%	Spinocerebellar ataxia 27, 609307
FLVCR1	105.2	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FTL	103.6	100%	99%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
GALC	104.6	100%	98%	Krabbe disease, 245200
GAN	149.0	100%	100%	Giant axonal neuropathy-1, 256850

GBA	71.0	63%	58%	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 {Parkinson disease, late-onset, susceptibility to}, 168600 {Lewy body dementia, susceptibility to}, 127750
GBA2	129.1	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	90.2	92%	92%	Glutaricaciduria, type I, 231670
GCH1	117.3	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	85.3	100%	97%	Alexander disease, 203450
GJC2	55.0	97%	87%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GLB1	76.8	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	108.1	100%	100%	Dystonia 25, 615073
GOSR2	102.6	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GPR56	102.2	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GRM1	156.8	100%	98%	Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	120.7	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	45.3	88%	69%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HSPD1	16.6	60%	44%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
ITPR1	111.4	99%	97%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	118.1	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNC3	85.7	72%	67%	Spinocerebellar ataxia 13, 605259
KCNJ10	170.0	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCTD7	113.5	93%	86%	Epilepsy,progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	103.8	100%	98%	Spastic paraplegia 8, autosomal dominant, 603563

KIAA0226	103.1	99%	98%	?Spinocerebellar ataxia, autosomal recessive 15
KIF1A	77.2	99%	95%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF1C	118.8	100%	97%	Spastic ataxia 2,autosomal recessive, 611302
KIF5A	99.0	99%	97%	Spastic paraplegia 10, autosomal dominant, 604187
L1CAM	65.4	99%	93%	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 agenesis of, 304100
MARS2	169.1	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MECP2	89.9	97%	88%	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 Mental retardation, X-linked syndromic, Lubs type, 300260
MICU1	98.8	100%	100%	Myopathy with extrapyramidal signs
MMADHC	72.3	100%	100%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MRE11A	99.1	100%	100%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	110.1	100%	99%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTPAP	117.1	91%	91%	Ataxia, spastic, 4, 613672
MTTP	106.2	98%	97%	Abetalipoproteinemia, 200100; {Metabolic syndrome, protection against}, 605552
NIPA1	100.5	97%	88%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	105.7	100%	97%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NOL3	133.7	100%	100%	Myoclonus, familial cortical, 614937

NPC1	97.6	100%	98%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	71.2	100%	100%	Niemann-pick disease, type C2, 607625
NUP62	94.0	100%	100%	Striatonigral degeneration, infantile, 271930
OPA1	135.7	100%	98%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
PANK2	126.3	100%	99%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX6	99.1	100%	99%	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 optic nerve, 120430 Gillespie syndrome, 206700
PDE8B	103.0	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDHA1	66.8	95%	91%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	108.2	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	102.2	87%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	94.5	100%	96%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	165.3	100%	100%	Spinocerebellar ataxia 23, 610245
PEX10	82.5	97%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX7	109.6	96%	92%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	93.5	100%	99%	Refsum disease, 266500
PIK3R5	82.1	100%	100%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	83.7	99%	91%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLP1	42.0	96%	84%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920

PMM2	101.0	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKD	53.1	98%	91%	Paroxysmal nonkinesigenic dyskinesia, 118800
PNPLA6	93.4	99%	97%	Spastic paraplegia 39, autosomal recessive, 612020
POLG	94.1	98%	95%	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), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459
PRKCG	109.3	98%	94%	Spinocerebellar ataxia 14, 605361
PRKRA	129.7	100%	100%	Dystonia 16, 612067
PRRT2	85.6	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PSEN1	105.9	100%	93%	Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694 Acne inversa, familial, 3, 613737
REEP1	102.7	100%	94%	Spastic paraplegia 31, autosomal dominant, 610250 Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	98.2	100%	93%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	106.4	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	148.5	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF170	125.4	100%	100%	taxia, sensory, 1, autosomal dominant, 608984
RTN2	73.6	98%	93%	Spastic paraplegia 12, autosomal dominant, 604805
SACS	168.6	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	122.7	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SCN8A	146.1	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306; Epileptic encephalopathy, early infantile, 13, 614558
SERAC1	98.6	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	165.3	100%	99%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	94.7	95%	94%	maternally imprinted Dystonia-11, myoclonic, 159900

SIL1	101.6	100%	98%	Marinesco-Sjogren syndrome, 248800
SLC16A2	61.1	99%	90%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	117.6	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	128.1	100%	100%	Episodic ataxia, type 6, 612656
SLC25A15	111.4	94%	84%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC2A1	102.2	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC30A10	144.5	100%	100%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	100.8	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC52A2	116.4	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC6A3	87.4	100%	98%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SLC9A1	115.8	100%	99%	No OMIM disease phenotype
SMPD1	108.4	100%	96%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	129.9	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SPAST	117.4	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	114.0	100%	98%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	127.7	100%	100%	Troyer syndrome, 275900
SPG21	113.3	100%	100%	Mast syndrome, 248900
SPG7	89.0	98%	88%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	74.2	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	106.7	100%	98%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	111.4	100%	100%	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	197.1	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	118.1	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TAF1	79.3	100%	99%	Dystonia-Parkinsonism, X-linked, 314250
TDP1	120.2	100%	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250

TECPR2	116.9	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TGM6	74.9	93%	90%	Spinocerebellar ataxia 35, 613908
TH	97.3	96%	90%	Segawa syndrome, recessive, 605407
THAP1	129.7	100%	100%	Dystonia 6, torsion, 602629
TIMM8A	35.0	99%	73%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMEM240	129.5	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM67	120.7	100%	98%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOR1A	146.6	99%	97%	Dystonia-1, torsion, 128100 Dystonia, early-onset atypical, with myoclonic features {Dystonia-1, modifier of}
TREX1	147.2	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TTBK2	129.8	100%	100%	Spinocerebellar ataxia 11, 604432
TTC19	79.4	81%	77%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	93.2	93%	82%	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	64.6	87%	77%	?Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
VAMP1	116.0	100%	99%	spastic ataxia 1, autosomal dominant
VCP	118.6	100%	96%	Spastic Ataxia 1, autosomal dominant, 108600
VLDLR	116.5	100%	98%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	127.2	100%	99%	Choreoacanthocytosis, 200150
VPS37A	85.5	98%	92%	Spastic paraplegia 53, autosomal recessive, 614898
WDR45	46.6	98%	89%	?Neurodegeneration with brain iron acculation 5, 300894
WDR81	120.4	99%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	113.4	100%	100%	Esophageal squamous cell carcinoma, 133239; Spinocerebellar ataxia 12, 614322
ZFYVE26	96.7	97%	93%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	87.5	100%	98%	Spastic paraplegia 33, autosomal dominant, 610244

ZNF592	122.4	94%	92%	Spinocerebellar ataxia, autosomal recessive 5, 606937

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
Median Coverage describes the average number of reads seen across 50 exomes
% 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
OMIM release used for OMIM disease identifiers and descriptions : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors