Whole Exome Sequencing
Gene package Neurodegeneration, version 4, 30-9-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. Additionally, MPLA analysis was performed for *APP* (SALSA P170 APP; MRC Holland) and several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). For *ATN1*, *C9orf72 and TBP* a repeat expansion test was performed. *PRNP* was also tested by Sanger sequencing. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
AAAS	Achalasia-addisonianism-alacrimia syndrome, 231550	605378	102	100	100	100
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889	612035	122	100	100	100
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	86	84	78	75
ADPRHL2	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170	610624	129	100	100	100
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276	606830	64	100	95	87
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225	606352	72	100	100	96
AMPD2	Pontocerebellar hypoplasia, type 9, 615809 ?Spastic paraplegia 63, 615686	102771	142	100	100	100
ANG	Amyotrophic lateral sclerosis 9, 611895	105850	126	100	100	100
ANXA11	Amytrophic lateral sclerosis 23, 617839	602572	80	100	100	98
APP	Alzheimer disease 1, familial, 104300 Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714	104760	85	100	100	99
ARHGEF28	No OMIM phenotype	612790	60	100	97	90
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ATP13A2	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, 617225	610513	117	100	100	98
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	144	100	100	100
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	46	100	91	64
ATP7B	Wilson disease, 277900	606882	99	100	100	99
AUH	3-methylglutaconic aciduria, type I, 250950	600529	94	100	100	98
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	186	100	100	100
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	614260	56	100	100	96
CACNA1A	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086	601011	87	100	98	93
CAV1	?Lipodystrophy, congenital generalized, type 3, 612526 Lipodystrophy, familial partial, type 7, 606721 Pulmonary hypertension, primary, 3, 615343	601047	95	100	100	100
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	127	100	100	100
CHCHD10	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, 616209 Spinal muscular atrophy, Jokela type, 615048	615903	34	86	65	52
CHCHD2	Parkinson disease 22, 616710	616244	67	100	88	68
CHMP2B	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795	609512	83	100	100	97
CISD2	Wolfram syndrome 2, 604928	611507	137	100	100	100
CLCN2	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651	600570	100	100	100	100
CLN3	Ceroid lipofuscinosis, neuronal, 3, 204200	607042	111	100	100	100
CLN5	Ceroid lipofuscinosis, neuronal, 5, 256731	608102	70	100	100	100
CLN6	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300	606725	120	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	129	100	100	100
COASY	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266	609855	139	100	100	100
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 {Hemorrhage, intracerebral, susceptibility to}, 614519 ?Retinal arteries, tortuosity of, 180000	120130	88	100	100	97
COL4A2	Brain small vessel disease 2, 614483 {Hemorrhage, intracerebral, susceptibility to}, 614519	120090	102	100	100	99
COQ4	Coenzyme Q10 deficiency, primary, 7, 616276	612898	110	100	100	100
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	63	100	100	96
CRAT	?Neurodegeneration with brain iron accumulation 8, 617917	600184	95	100	100	99
CSF1R	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	99	100	100	100
CST3	Cerebral amyloid angiopathy, 105150 {Macular degeneration, age-related, 11}, 611953	604312	97	100	100	92
CTSA	Galactosialidosis, 256540	613111	124	100	100	100
CTSD	Ceroid lipofuscinosis, neuronal, 10, 610127	116840	137	100	100	100
CTSF	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362	603539	94	100	94	91
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	131	100	100	100
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105	610956	63	100	100	97
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	75	100	100	94
DCTN1	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	601143	95	100	100	99
DNAJC13	No OMIM phenotype	614334	71	100	99	94
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350	611203	217	100	100	100
DNAJC6	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	608375	84	100	100	98
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	93	100	100	97
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896	606686	79	100	100	100
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606454	77	100	98	91
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896	606273	58	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
EIF2B4	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606687	89	100	100	100
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	603945	72	100	100	98
EIF4G1	{Parkinson disease 18}, 614251	600495	100	100	100	100
ERBB4	Amyotrophic lateral sclerosis 19, 615515	600543	65	100	99	93
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	114	100	100	96
FA2H	Spastic paraplegia 35, 612319	611026	83	100	99	91
FBXO7	Parkinson disease 15, 260300	605648	86	100	100	97
FIG4	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 ?Polymicrogyria, bilateral temporooccipital, 612691 Yunis-Varon syndrome, 216340	609390	59	100	99	94
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	111	100	100	100
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	144	100	100	100
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782	137070	77	100	100	97
GALC	Krabbe disease, 245200	606890	54	100	99	89
GBE1	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570	607839	67	100	99	92
GCDH	Glutaricaciduria, type I, 231670	608801	112	100	100	100
GFAP	Alexander disease, 203450	137780	81	100	100	99
GIGYF2	{Parkinson disease 11}, 607688	612003	66	100	99	94
GLA	Fabry disease, 301500 Fabry disease, cardiac variant, 301500	300644	101	100	100	100
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	155	100	100	100
GSN	Amyloidosis, Finnish type, 105120	137350	95	100	100	99
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	86	100	100	98
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	119	100	100	97
HNRNPA1	Amyotrophic lateral sclerosis 20, 615426 ?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424	164017	61	100	99	95
HNRNPA2B1	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422	600124	58	100	100	95

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
HTRA1	CARASIL syndrome, 600142 Cerebral arteriopathy, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149	602194	93	96	87	83
HTRA2	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297	606441	143	100	100	98
ISCA2	Multiple mitochondrial dysfunctions syndrome 4, 616370	615317	89	100	100	100
ITM2B	Dementia, familial British, 176500 Dementia, familial Danish, 117300 ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079	603904	79	100	100	97
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	159	100	100	100
LMNB1	Leukodystrophy, adult-onset, 169500	150340	66	100	100	97
LRP10	No OMIM phenotype	609921	148	100	100	100
LRRK2	{Parkinson disease 8}, 607060	609007	78	100	100	96
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540	157140	120	100	100	100
MATR3	Amyotrophic lateral sclerosis 21, 606070	164015	63	100	100	95
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	97	100	100	99
MED20	No OMIM phenotype	612915	148	100	100	100
MFSD8	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170	611124	68	100	100	95
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type, 277400	609831	133	100	100	100
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	99	100	100	99
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 {Neural tube defects, folate-sensitive, susceptibility to}, 601634	156570	80	100	100	97
MYORG	Basal ganglia calcification, idiopathic, 7, 618317	618255	205	100	100	99
NKX6-2	Spastic ataxia 8, with hypomyelinating leukodystrophy, 617560	605955	107	99	88	83
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 Lateral meningocele syndrome, 130720 ?Myofibromatosis, infantile 2, 615293	600276	111	99	95	91
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	87	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
NPC2	Niemann-pick disease, type C2, 607625	601015	95	100	100	100
OPTN	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657	602432	81	100	100	96
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	68	100	99	95
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	81	100	100	99
PARK7	Parkinson disease 7 early-onset, 606324	602533	68	100	99	87
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174	190040	101	100	100	100
PDGFRB	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812	173410	116	100	100	100
PFN1	Amyotrophic lateral sclerosis 18, 614808	176610	159	100	100	100
PINK1	Parkinson disease 6, early onset, 605909	608309	100	98	93	88
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	118	100	100	99
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	114	100	100	99
PPT1	Ceroid lipofuscinosis, neuronal, 1, 256730	600722	84	100	100	100
PRKAR1B	No OMIM phenotype	176911	95	100	98	91
PRKN	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116	602544	96	100	100	99
PRKRA	Dystonia 16, 612067	603424	81	100	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PRNP	Cerebral amyloid angiopathy, PRNP-related, 137440 Creutzfeldt-Jakob disease, 123400 Gerstmann-Straussler disease, 137440 Huntington disease-like 1, 603218 Insomnia, fatal familial, 600072 {Kuru, susceptibility to}, 245300 Prion disease with protracted course, 606688	176640	138	100	100	100
PSEN1	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700	104311	59	100	100	93
PSEN2	Alzheimer disease-4, 606889 Cardiomyopathy, dilated, 1V, 613697	600759	113	100	100	100
PSENEN	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736	607632	88	100	100	100
PTRHD1	No OMIM phenotype	617342	119	100	100	100
QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760	603727	108	100	100	100
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	66	100	99	93
REPS1	?Neurodegeneration with brain iron accumulation 7, 617916	614825	54	98	94	88
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	78	100	100	96
SERPINI1	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218	602445	73	100	100	98
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, with axonal neuropathy 2, 606002	608465	70	100	100	97
SIGMAR1	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, 2, 605726	601978	135	100	100	100
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	86	100	100	97
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	99	100	100	95
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	68	100	98	86
SLC39A14	Hypermanganesemia with dystonia 2, 617013 ?Hyperostosis cranalis interna, 144755	608736	92	100	100	97
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135	126455	102	100	100	100
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	131	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SNCA	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	163890	52	100	100	98
SNCB	Dementia, Lewy body, 127750	602569	130	100	100	100
SNORD118	Leukoencephalopathy, brain calcifications, and cysts, 614561	616663	No coverage data			
SOD1	Amyotrophic lateral sclerosis 1, 105400	147450	74	100	100	100
SORL1	No OMIM phenotype	602005	86	100	100	96
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	84	100	100	97
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	102	100	100	100
SQSTM1	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250	601530	129	100	100	96
STUB1	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia 16, 615768	607207	137	100	100	100
SUMF1	Multiple sulfatase deficiency, 272200	607939	88	100	100	99
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	62	100	99	92
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	54	100	97	87
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	96	100	100	99
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069	605078	124	100	100	95
TBC1D23	Pontocerebellar hypoplasia, type 11, 617695	617687	57	100	99	90
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	104	100	99	96
TBK1	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439	604834	80	100	99	90
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	80	100	100	96
TH	Segawa syndrome, recessive, 605407	191290	93	100	99	94
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	110	100	100	98
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	97	100	100	100
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193	605086	107	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	234	100	100	100
TSEN2	Pontocerebellar hypoplasia type 2B, 612389	608753	63	87	86	78
TTR	Amyloidosis, hereditary, transthyretin-related, 105210 Carpal tunnel syndrome, familial, 115430 [Dystransthyretinemic hyperthyroxinemia], 145680	176300	93	100	100	100
TUBA4A	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208	191110	200	100	100	100
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041	131222	102	100	100	100
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770	604142	108	100	100	100
UBQLN2	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857	300264	79	100	100	100
UCHL1	{?Parkinson disease 5, susceptibility to}, 613643 Spastic paraplegia 79, 615491	191342	79	100	100	99
VAPB	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980	605704	71	100	100	98
VCP	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320	601023	90	100	100	98
VPS13A	Choreoacanthocytosis, 200150	605978	72	100	99	91
VPS13C	Parkinson disease 23, early onset, 616840	608879	50	100	97	85
VPS35	{Parkinson disease 17}, 614203	601501	68	100	99	94
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	82	100	100	96
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	92	100	100	100
XPR1	Basal ganglia calcification, idiopathic, 6, 616413	605237	64	100	100	99

- Gene symbols according HGCN
- OMIM release used: 8-9-2919
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x