Whole Exome Sequencing
Gene package Neurodegeneration, version 5, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique 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 Alissa Interpret 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AAAS	605378	175	100	100	100
AARS2	612035	239	100	100	100
ABCD1	300371	178	100	100	100
ADPRHL2	610624	176	100	100	100
AGTPBP1	606830	63	95	86	73
ALS2	606352	120	100	98	96
AMPD2	102771	219	100	100	100
ANG	105850	136	100	100	100
ANXA11	602572	180	100	100	100
APP	104760	128	100	100	99
ARHGEF28	612790	99	98	95	90
ARSA	607574	380	100	100	100
ATP13A2	610513	228	100	100	100
ATP1A3	182350	221	100	100	100
ATP6AP2	300556	54	100	93	81
ATP7B	606882	195	100	100	100
AUH	600529	81	100	98	91
C19orf12	614297	235	100	100	100
C9orf72	614260	88	100	100	97
CACNA1A	601011	139	100	100	99
CAV1	601047	134	100	100	100
CBS	613381	430	100	100	100
CHCHD10	615903	135	95	93	92

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CHCHD2	616244	171	100	100	98
CHMP2B	609512	44	85	75	54
CISD2	611507	97	76	76	76
CLCN2	600570	275	100	100	100
CLN3	607042	211	100	100	100
CLN5	608102	113	100	93	89
CLN6	606725	264	100	100	100
CLN8	607837	195	100	100	100
COASY	609855	239	100	100	100
COL4A1	120130	121	100	99	96
COL4A2	120090	196	100	100	99
COQ4	612898	176	100	100	97
CP	117700	92	99	98	94
CRAT	600184	239	100	100	100
CSF1R	164770	205	100	100	100
CST3	604312	186	100	100	100
CTSA	613111	181	100	100	100
CTSD	116840	278	100	100	100
CTSF	603539	232	100	98	96
CYP27A1	606530	295	100	100	100
DARS2	610956	79	100	96	94
DCAF17	612515	71	100	99	93
DCTN1	601143	162	100	100	99
DNAJC13	614334	58	99	91	78
DNAJC5	611203	258	100	100	100
DNAJC6	608375	125	100	100	96
DNMT1	126375	192	100	100	98
EIF2B1	606686	126	100	100	99
EIF2B2	606454	152	100	100	99
EIF2B3	606273	121	100	100	99
EIF2B4	606687	177	100	98	93
EIF2B5	603945	146	100	100	100
EIF4G1	600495	178	100	100	99
ERBB4	600543	112	100	100	97
EXOSC3	606489	208	100	100	100
FA2H	611026	182	100	100	100
FBXO7	605648	126	97	94	90
FIG4	609390	88	100	96	91
FOLR1	136430	146	100	100	100
FTL	134790	188	100	100	100
FUS	137070	135	100	100	100
GALC	606890	85	98	94	88
GBE1	607839	87	100	100	98

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GCDH	608801	239	100	100	100
GFAP	137780	176	100	100	100
GIGYF2	612003	80	100	98	91
GLA	300644	87	100	100	98
GRN	138945	336	100	100	100
GSN	137350	201	100	100	100
HEXA	606869	162	100	100	100
HEXB	606873	73	97	91	84
HNRNPA1	164017	114	100	99	94
HNRNPA2B1	600124	69	100	99	90
HTRA1	602194	144	99	91	89
HTRA2	606441	256	100	100	100
ISCA2	615317	177	100	100	100
ITM2B	603904	87	100	96	88
JAM2	606870	65	100	96	87
KCTD7	611725	259	100	100	100
LMNB1	150340	110	100	97	92
LRP10	609921	294	100	100	100
LRRK2	609007	65	98	92	84
MAPT	157140	170	100	100	99
MATR3	164015	86	98	92	85
MECR	608205	137	100	100	99
MED20	612915	168	100	100	100
MFSD8	611124	82	99	93	83
MMACHC	609831	205	100	100	100
MTHFR	607093	191	100	100	100
MTR	156570	119	100	100	98
MYORG	618255	583	100	100	100
NKX6-2	605955	203	100	100	100
NOTCH3	600276	291	100	100	99
NOVA2	601991	210	99	96	94
NPC1	607623	138	100	100	99
NPC2	601015	119	100	100	99
NRROS	615322	318	100	100	100
OPTN	602432	81	99	90	77
PAH	612349	121	100	100	99
PANK2	606157	117	100	99	97
PARK7	602533	76	100	99	85
PDGFB	190040	184	100	100	100
PDGFRB	173410	235	100	100	100
PFN1	176610	190	100	100	100
PINK1	608309	258	100	100	98
PLA2G6	603604	287	100	100	100

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POLG	174763	188	100	100	100
PPT1	600722	145	100	100	100
PRKAR1B	176911	180	100	100	100
PRKN	602544	138	100	100	99
PRKRA	603424	101	100	100	99
PRNP	176640	470	100	100	100
PSEN1	104311	130	100	99	92
PSEN2	600759	181	100	100	100
PSENEN	607632	150	100	100	100
PTRHD1	617342	264	100	100	100
QARS1	603727	206	100	100	100
RARS2	611524	70	100	93	77
REPS1	614825	89	100	95	89
SEMA6B	608873	199	100	100	100
SEPSECS	613009	88	99	96	90
SERPINI1	602445	58	100	98	89
SETX	608465	100	100	97	95
SIGMAR1	601978	318	100	100	100
SLC17A5	604322	82	100	94	85
SLC20A2	158378	185	100	95	95
SLC33A1	603690	147	98	93	85
SLC39A14	608736	159	100	100	100
SLC44A1	606105	74	98	94	87
SLC5A6	604024	199	100	100	100
SLC6A3	126455	226	100	100	100
SMPD1	607608	377	100	100	100
SNCA	163890	106	100	100	100
SNCB	602569	155	100	100	100
SNORD118	616663	No coverage data			
SOD1	147450	154	100	100	100
SORL1	602005	143	100	100	99
SPG11	610844	105	99	94	88
SPR	182125	142	100	100	100
SQSTM1	601530	268	100	100	100
STUB1	607207	435	100	100	100
SUMF1	607939	174	100	100	100
SYNJ1	604297	83	98	95	90
TAF1	313650	71	100	99	95
TANGO2	616830	253	100	100	100
TARDBP	605078	152	100	100	97
TBC1D23	617687	64	91	87	77
TBCD	604649	210	99	97	95
TBK1	604834	52	93	79	65

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TBP	600075	140	100	99	90
TH	191290	266	100	100	100
TOE1	613931	204	100	100	100
TPP1	607998	223	100	100	100
TREM2	605086	225	100	100	100
TREX1	606609	313	100	100	100
TSEN2	608753	190	87	87	84
TTR	176300	168	100	100	100
TUBA4A	191110	221	100	100	100
TYMP	131222	204	100	100	100
TYROBP	604142	161	100	100	100
UBQLN2	300264	116	100	100	100
UCHL1	191342	89	100	94	84
VAPB	605704	71	100	96	82
VCP	601023	140	100	100	100
VPS13A	605978	48	95	81	63
VPS13C	608879	63	96	87	77
VPS35	601501	82	100	97	91
VPS53	615850	133	100	100	100
WDR45	300526	161	100	100	100
XPR1	605237	87	100	98	94

- OMIM release used: 8-9-2019
- 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