Whole Exome Sequencing
Gene package Movement disorders, version 8, 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 *APTX, FXN* and *SETX* (SALSA P316 Recessive Ataxias probemix; MRC Holland) and for several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). For *ATN1*, *ATXN1, ATXN2, ATXN3, ATXN7, C9orf72, FMR1, PPP2R2B* and *TBP* a repeat expansion test was performed. For *FXN* and *CACNA1A* a repeat expansion test was performed in additional to either MPLA and/or exome 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
AARS1	601065	161	100	100	100
AARS2	612035	239	100	100	100
ABCA2	600047	329	100	100	100
ABCB7	300135	64	100	94	83
ABCD1	300371	178	100	100	100
ABHD12	613599	133	100	99	93
ACTB	102630	253	100	100	100
ADAR	146920	163	100	100	100
ADCY5	600293	259	100	99	98
ADGRG1	604110	233	100	100	100
ADPRHL2	610624	176	100	100	100
AFG3L2	604581	107	96	96	93
AGTPBP1	606830	63	95	86	73
AIMP1	603605	70	99	88	81
ALDH18A1	138250	169	100	100	99
ALDH3A2	609523	90	100	100	97
ALDH5A1	610045	121	100	100	98
ALS2	606352	120	100	98	96
ANO10	613726	82	98	93	85
ANO3	610110	61	100	98	90
AP4B1	607245	130	100	100	100
AP4E1	607244	66	97	87	75
AP4M1	602296	141	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AP4S1	607243	50	100	96	78
AP5Z1	613653	253	100	100	100
APTX	606350	166	100	98	90
ARG1	608313	89	100	99	95
ARL6IP1	607669	71	91	81	69
ARSA	607574	380	100	100	100
ARX	300382	100	92	89	84
ASPA	608034	103	100	98	94
ATCAY	608179	200	100	100	100
ATG5	604261	51	93	81	74
ATL1	606439	101	98	98	93
ATM	607585	61	96	86	75
ATN1	607462	189	100	100	100
ATP13A2	610513	228	100	100	100
ATP1A2	182340	189	100	100	100
ATP1A3	182350	221	100	100	100
ATP2B3	300014	160	100	100	99
ATP6AP2	300556	54	100	93	81
ATP7B	606882	195	100	100	100
ATP8A2	605870	125	98	96	94
ATXN1	601556	263	100	100	100
ATXN2	601517	92	91	82	75
ATXN3	607047	75	93	92	83
ATXN7	607640	116	99	96	94
AUH	600529	81	100	98	91
B4GALNT1	601873	269	100	100	100
BCAP31	300398	82	100	100	95
BCKDHA	608348	221	100	100	100
BCKDHB	248611	137	100	100	99
BSCL2	606158	178	100	100	100
C12orf65	613541	100	100	94	58
C19orf12	614297	235	100	100	100
C9orf72	614260	88	100	100	97
CA8	114815	95	100	100	94
CACNA1A	601011	139	100	100	99
CACNA1B	601012	200	98	97	96
CACNA1E	601013	174	100	100	100
CACNA1G	604065	219	100	100	100
CACNB4	601949	176	100	100	99
CAMTA1	611501	210	100	98	95
CAPN1	114220	297	100	100	100
CCDC88C	611204	187	100	100	98
CCT5	610150	135	100	99	92
CDKL5	300203	74	97	92	86

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CHCHD2	616244	171	100	100	98
CHMP1A	164010	202	100	100	100
CHP1	606988	136	100	100	100
CIZ1	611420	162	100	100	100
CLCN2	600570	275	100	100	100
CLCN4	302910	141	100	100	98
CLP1	608757	213	100	100	100
CLPB	616254	209	100	100	100
COASY	609855	239	100	100	100
COL6A3	120250	210	100	100	99
COQ2	609825	79	98	92	82
COQ8A	606980	250	100	100	100
COQ9	612837	123	100	100	100
CP	117700	92	99	98	94
CPT1C	608846	176	100	100	100
CSF1R	164770	205	100	100	100
CSTB	601145	132	100	100	100
CWF19L1	616120	91	100	94	88
CYB5R3	613213	188	100	100	100
CYP27A1	606530	295	100	100	100
CYP2U1	610670	143	100	100	99
CYP7B1	603711	111	100	99	94
DAB1	603448	135	100	98	93
DBT	248610	70	100	93	83
DCAF17	612515	71	100	99	93
DCC	120470	114	100	100	98
DCTN1	601143	162	100	100	99
DDC	107930	112	100	95	92
DDHD1	614603	143	100	100	97
DDHD2	615003	71	100	94	87
DHDDS	608172	146	100	100	100
DLAT	608770	131	100	99	92
DLD	238331	65	99	90	76
DNAJC13	614334	58	99	91	78
DNAJC3	601184	68	97	85	80
DNAJC6	608375	125	100	100	96
DNAL4	610565	119	100	100	100
DNMT1	126375	192	100	100	98
DSTYK	612666	167	100	100	100
ECHS1	602292	170	100	100	100
EEF2	130610	296	100	100	100
EIF2AK2	176871	46	93	74	59
EIF2B1	606686	126	100	100	99
EIF2B2	606454	152	100	100	99

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EIF2B3	606273	121	100	100	99
EIF2B4	606687	177	100	98	93
EIF2B5	603945	146	100	100	100
EIF4G1	600495	178	100	100	99
ELOVL4	605512	98	98	89	81
ELOVL5	611805	105	100	100	98
ENTPD1	601752	102	100	100	99
ERLIN1	611604	87	100	100	99
ERLIN2	611605	110	100	100	100
EXOSC3	606489	208	100	100	100
FA2H	611026	182	100	100	100
FAM126A	610531	96	100	100	94
FAR1	616107	69	100	92	82
FARS2	611592	221	100	100	100
FAT2	604269	229	100	100	100
FBXO7	605648	126	97	94	90
FGF14	601515	93	100	97	89
FLVCR1	609144	132	100	95	85
FMR1	309550	39	96	84	63
FOLR1	136430	146	100	100	100
FOXG1	164874	424	100	97	94
FRMD7	300628	80	100	99	93
FTL	134790	188	100	100	100
FUS	137070	135	100	100	100
FXN	606829	96	100	100	99
GALC	606890	85	98	94	88
GAN	605379	123	100	99	96
GBA	606463	207	100	100	100
GBA2	609471	212	100	100	100
GCDH	608801	239	100	100	100
GCH1	600225	207	94	94	94
GFAP	137780	176	100	100	100
GIGYF2	612003	80	100	98	91
GJC2	608803	283	100	100	98
GLB1	611458	149	100	100	100
GNAL	139312	114	100	100	99
GOSR2	604027	139	100	100	100
GPR143	300808	111	100	100	99
GRID2	602368	117	100	100	98
GRIN1	138249	259	100	100	100
GRIN2B	138252	237	100	100	99
GRM1	604473	210	100	100	99
HACE1	610876	59	97	89	77
HEXB	606873	73	97	91	84

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HPCA	142622	250	100	100	100
HPRT1	308000	40	95	80	55
HSD17B4	601860	78	93	91	84
HSPD1	118190	67	99	90	77
HTRA2	606441	256	100	100	100
IBA57	615316	227	100	100	100
IFRD1	603502	66	98	90	78
ISCA2	615317	177	100	100	100
ITPR1	147265	120	100	99	97
JAM2	606870	65	100	96	87
KATNB1	602703	334	100	100	100
KCNA1	176260	224	100	100	100
KCNA2	176262	209	100	100	100
KCNC1	176258	275	100	100	100
KCNC3	176264	321	94	88	87
KCND3	605411	350	100	100	99
KCNJ10	602208	271	100	100	100
KCNMA1	600150	122	100	98	96
KCTD17	616386	156	100	100	100
KCTD7	611725	259	100	100	100
KIDINS220	615759	106	100	98	93
KIF1A	601255	260	100	100	100
KIF1C	603060	225	100	100	100
KIF5A	602821	126	100	100	100
KMT2B	606834	247	99	98	97
L1CAM	308840	164	100	100	100
LAMA1	150320	140	100	99	98
LAMB1	150240	144	100	98	94
LMNB1	150340	110	100	97	92
LRP10	609921	294	100	100	100
LRPPRC	607544	61	98	90	79
LRRK2	609007	65	98	92	84
MAG	159460	302	100	100	100
MAPT	157140	170	100	100	99
MARS2	609728	343	100	100	100
MECP2	300005	200	100	100	99
MECR	608205	137	100	100	99
MED20	612915	168	100	100	100
MICU1	605084	88	98	92	84
MLC1	605908	181	100	100	95
MMADHC	611935	56	85	85	80
MME	120520	53	92	79	67
MRE11	600814	61	97	88	76
MTHFR	607093	191	100	100	100

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MTPAP	613669	69	99	89	77
MTTP	157147	89	100	100	98
MVK	251170	207	100	100	100
MYBPC1	160794	90	100	99	95
MYORG	618255	583	100	100	100
NEFL	162280	244	100	99	95
NEU1	608272	238	100	100	100
NEXMIF	300524	94	100	100	100
NFASC	609145	192	100	100	100
NIPA1	608145	158	100	100	99
NKX2-1	600635	216	100	100	100
NKX6-2	605955	203	100	100	100
NOL3	605235	314	100	100	100
NOTCH2NLC	618025	No coverage data			
NPC1	607623	138	100	100	99
NPC2	601015	119	100	100	99
NT5C2	600417	78	95	90	84
NUP62	605815	345	100	100	100
OCLN	602876	100	85	81	69
OPA1	605290	55	90	79	67
OPHN1	300127	71	100	99	91
PANK2	606157	117	100	99	97
PARK7	602533	76	100	99	85
PAX6	607108	267	100	100	100
PCNA	176740	143	100	96	86
PCYT2	602679	230	100	100	98
PDE10A	610652	94	97	97	95
PDE8B	603390	100	100	100	97
PDGFB	190040	184	100	100	100
PDGFRB	173410	235	100	100	100
PDHA1	300502	78	100	94	87
PDHX	608769	79	100	95	89
PDSS1	607429	69	100	96	82
PDSS2	610564	152	99	93	88
PDYN	131340	181	100	100	100
PEX10	602859	266	100	100	100
PEX2	170993	93	100	100	100
PEX7	601757	94	100	100	94
PHYH	602026	99	100	97	87
PIK3R5	611317	219	100	100	100
PINK1	608309	258	100	100	98
PLA2G6	603604	287	100	100	100
PLD3	615698	219	100	100	100
PLP1	300401	86	100	98	92

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PMM2	601785	115	100	100	99
PMPCA	613036	189	100	100	100
PNKD	609023	198	100	100	100
PNKP	605610	246	100	100	100
PNPLA6	603197	269	100	100	100
POLG	174763	188	100	100	100
POLR1C	610060	136	100	100	100
POLR3A	614258	143	100	100	99
POLR3B	614366	93	100	97	92
PPP2R2B	604325	108	100	100	99
PRF1	170280	383	100	100	100
PRICKLE1	608500	107	100	100	99
PRKCG	176980	212	100	100	100
PRKN	602544	138	100	100	99
PRKRA	603424	101	100	100	99
PRRT2	614386	179	100	100	98
PSEN1	104311	130	100	99	92
PTRHD1	617342	264	100	100	100
PUM1	607204	145	100	100	100
PYCR2	616406	201	100	100	100
RAB18	602207	68	91	90	85
RAB29	603949	102	100	100	100
RAB3GAP1	602536	86	100	99	95
RAB3GAP2	609275	96	97	91	81
RAD51	179617	102	88	88	88
RARS1	107820	75	100	98	91
RARS2	611524	70	100	93	77
REEP1	609139	164	100	100	98
RETREG1	613114	112	96	89	85
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	100	100	100
RNF170	614649	64	100	91	76
RNF216	609948	149	100	98	97
ROBO3	608630	213	100	100	100
RTN2	603183	218	100	100	100
RUBCN	613516	163	100	100	100
SACS	604490	97	100	100	98
SAMD9L	611170	85	100	100	99
SAMHD1	606754	97	100	99	93
SCN11A	604385	100	100	99	94
SCN2A	182390	93	98	94	86
SCN8A	600702	137	100	100	98
SCYL1	607982	194	100	100	100

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SEMA6B	608873	199	100	100	100
SEPSECS	613009	88	99	96	90
SERAC1	614725	98	98	95	89
SETX	608465	100	100	97	95
SGCE	604149	104	97	86	82
SIL1	608005	202	100	100	98
SLC12A6	604878	98	100	97	95
SLC16A2	300095	101	100	100	100
SLC17A5	604322	82	100	94	85
SLC19A3	606152	108	100	100	100
SLC1A3	600111	106	100	99	95
SLC20A2	158378	185	100	95	95
SLC25A15	603861	152	100	100	100
SLC2A1	138140	354	100	100	100
SLC30A10	611146	209	100	100	100
SLC33A1	603690	147	98	93	85
SLC39A14	608736	159	100	100	100
SLC52A2	607882	210	100	100	100
SLC52A3	613350	271	100	100	100
SLC6A19	608893	229	100	100	100
SLC6A3	126455	226	100	100	100
SLC9A1	107310	267	100	100	100
SLC9A6	300231	89	98	88	81
SMPD1	607608	377	100	100	100
SNCA	163890	106	100	100	100
SNORD118	616663	No coverage data			
SNX14	616105	43	83	65	53
SOX10	602229	381	100	100	100
SPART	607111	96	100	98	95
SPAST	604277	82	99	84	69
SPG11	610844	105	99	94	88
SPG21	608181	87	100	98	90
SPG7	602783	227	100	100	100
SPR	182125	142	100	100	100
SPTBN2	604985	211	100	99	98
STUB1	607207	435	100	100	100
SUCLA2	603921	74	100	98	88
SUCLG1	611224	90	100	100	96
SUMF1	607939	174	100	100	100
SUOX	606887	254	100	100	100
SURF1	185620	158	100	98	95
SYNE1	608441	113	100	98	95
SYNJ1	604297	83	98	95	90
SYT14	610949	71	100	94	82

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TAF1	313650	71	100	99	95
TANGO2	616830	253	100	100	100
TBC1D20	611663	149	100	100	96
TBC1D23	617687	64	91	87	77
TBCD	604649	210	99	97	95
TBP	600075	140	100	99	90
TDP1	607198	104	100	99	93
TDP2	605764	121	89	88	84
TECPR2	615000	195	100	99	97
TENM4	610084	205	100	100	100
TFG	602498	86	99	89	83
TGM6	613900	265	100	100	100
TH	191290	266	100	100	100
THAP1	609520	115	100	100	96
THG1L	618802	88	100	98	85
TIMM8A	300356	98	100	100	100
TMEM106B	613413	59	92	85	74
TMEM230	617019	91	81	81	75
TMEM240	616101	287	100	100	100
TMEM67	609884	61	90	74	62
TOE1	613931	204	100	100	100
TOR1A	605204	157	100	100	100
TPP1	607998	223	100	100	100
TRAPPC4	610971	163	100	100	100
TREM2	605086	225	100	100	100
TREX1	606609	313	100	100	100
TRPC3	602345	211	97	92	84
TSEN2	608753	190	87	87	84
TSEN54	608755	201	100	100	98
TTBK2	611695	141	100	100	100
TTC19	613814	101	100	98	93
TTPA	600415	93	100	100	97
TUBA1A	602529	219	100	100	100
TUBB4A	602662	233	100	100	100
TUBG1	191135	334	100	100	100
TWNK	606075	231	100	100	100
TYROBP	604142	161	100	100	100
UBA5	610552	52	93	78	66
UBAP1	609787	131	100	100	98
UBQLN2	300264	116	100	100	100
UCHL1	191342	89	100	94	84
UGDH	603370	88	99	91	84
VAMP1	185880	139	100	100	92
VAMP2	185881	152	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
VCP	601023	140	100	100	100
VLDLR	192977	120	100	100	98
VPS13A	605978	48	95	81	63
VPS13C	608879	63	96	87	77
VPS13D	608877	115	100	100	99
VPS35	601501	82	100	97	91
VPS37A	609927	61	100	92	80
VPS53	615850	133	100	100	100
VRK1	602168	56	100	89	72
VWA3B	614884	101	100	99	94
WASHC5	610657	82	100	94	87
WDR26	617424	80	100	96	92
WDR45	300526	161	100	100	100
WDR73	616144	194	100	100	100
WDR81	614218	293	100	100	100
WFS1	606201	268	100	100	100
WWOX	605131	128	100	94	94
XK	314850	107	100	100	100
XPR1	605237	87	100	98	94
XRCC1	194360	200	100	100	100
ZC4H2	300897	88	100	100	100
ZFYVE26	612012	176	100	100	99
ZFYVE27	610243	213	100	100	100

- 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