Whole Exome Sequencing
Gene package Movement disorders, version 7, 21-2-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + 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 Cartagenia 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	102	100	100	100
AARS1	601065	89	100	100	96
AARS2	612035	122	100	100	100
ABCB7	300135	59	100	98	87
ABCD1	300371	86	84	78	75
ABHD12	613599	61	100	100	94
ACTB	102630	192	100	100	100
ADAR	146920	90	100	100	100
ADCY5	600293	111	98	95	93
ADGRG1	604110	120	100	100	100
ADPRHL2	610624	129	100	100	100
AFG3L2	604581	77	99	95	90
AGTPBP1	606830	64	100	95	87
AIMP1	603605	78	100	100	96
ALDH18A1	138250	80	100	100	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ALDH3A2	609523	64	100	99	93
ALDH5A1	610045	66	100	97	91
ALS2	606352	72	100	100	96
ANO10	613726	57	100	99	88
ANO3	610110	61	93	92	87
AP4B1	607245	81	100	100	100
AP4E1	607244	64	100	100	96
AP4M1	602296	130	100	100	99
AP4S1	607243	44	100	99	87
AP5Z1	613653	111	100	100	99
APTX	606350	76	100	99	92
ARG1	608313	75	100	100	98
ARL6IP1	607669	87	100	100	99
ARSA	607574	136	100	100	100
ARX	300382	46	89	79	68
ASPA	608034	55	100	99	91
ATCAY	608179	116	100	100	96
ATG5	604261	47	100	97	81
ATL1	606439	72	100	100	93
ATM	607585	75	100	99	95
ATN1	607462	155	100	100	99
ATP13A2	610513	117	100	100	98
ATP1A2	182340	130	100	100	100
ATP1A3	182350	144	100	100	100
ATP2B3	300014	96	100	100	99
ATP6AP2	300556	46	100	91	64
ATP7B	606882	99	100	100	99
ATP8A2	605870	69	100	99	94
ATXN1	601556	116	100	100	100
ATXN2	601517	75	97	93	86
ATXN3	607047	87	100	99	92
ATXN7	607640	90	100	97	93
AUH	600529	94	100	100	98
B4GALNT1	601873	111	100	100	100
BCAP31	300398	83	100	100	97
BCKDHA	608348	160	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
BCKDHB	248611	64	100	99	94
BSCL2	606158	101	100	100	100
C12orf65	613541	78	100	100	100
C19orf12	614297	186	100	100	100
C9orf72	614260	56	100	100	96
CA8	114815	79	100	100	94
CACNA1A	601011	87	100	98	93
CACNA1B	601012	116	99	97	96
CACNA1E	601013	106	100	99	97
CACNA1G	604065	146	100	100	100
CACNB4	601949	57	100	100	94
CAMTA1	611501	161	100	99	97
CAPN1	114220	135	100	100	100
CCDC88C	611204	104	100	100	97
CCT5	610150	85	100	100	91
CDKL5	300203	58	100	97	87
CHCHD2	616244	67	100	88	68
CHMP1A	164010	102	100	100	100
CHP1	606988	40	99	84	66
CIZ1	611420	100	99	95	91
CLCN2	600570	100	100	100	100
CLCN4	302910	80	100	100	98
CLP1	608757	99	100	100	100
CLPB	616254	114	100	100	100
COASY	609855	139	100	100	100
COL6A3	120250	104	100	100	99
COQ2	609825	66	100	100	94
COQ8A	606980	129	100	100	100
COQ9	612837	105	100	100	100
CP	117700	63	100	100	96
CPT1C	608846	107	100	100	98
CSF1R	164770	99	100	100	100
CSTB	601145	84	100	100	100
CWF19L1	616120	59	100	100	92
CYB5R3	613213	127	100	100	100
CYP27A1	606530	131	100	100	100

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CYP2U1	610670	66	100	100	96
CYP7B1	603711	67	100	100	98
DAB1	603448	70	100	100	97
DBT	248610	115	100	100	99
DCAF17	612515	75	100	100	94
DCC	120470	70	100	100	98
DCTN1	601143	95	100	100	99
DDC	107930	84	100	99	92
DDHD1	614603	107	100	98	92
DDHD2	615003	76	100	100	97
DHDDS	608172	69	100	100	99
DLAT	608770	74	100	98	90
DLD	238331	77	100	100	98
DNAJC13	614334	71	100	99	94
DNAJC3	601184	66	100	99	91
DNAJC6	608375	84	100	100	98
DNAL4	610565	121	100	100	100
DNMT1	126375	93	100	100	97
DSTYK	612666	77	100	98	93
ECHS1	602292	79	100	100	100
EEF2	130610	117	100	100	100
EIF2B1	606686	79	100	100	100
EIF2B2	606454	77	100	98	91
EIF2B3	606273	58	100	100	94
EIF2B4	606687	89	100	100	100
EIF2B5	603945	72	100	100	98
EIF4G1	600495	100	100	100	100
ELOVL4	605512	71	100	100	98
ELOVL5	611805	93	100	100	98
ENTPD1	601752	68	100	100	96
ERLIN1	611604	66	100	100	95
ERLIN2	611605	65	100	100	95
EXOSC3	606489	114	100	100	96
FA2H	611026	83	100	99	91
FAM126A	610531	63	100	100	98
FAR1	616107	74	100	100	96

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FARS2	611592	111	100	100	100
FAT2	604269	95	100	100	99
FBXO7	605648	86	100	100	97
FGF14	601515	68	100	100	98
FLVCR1	609144	98	100	100	96
FMR1	309550	41	100	96	76
FOLR1	136430	111	100	100	100
FOXG1	164874	127	97	90	84
FRMD7	300628	50	100	98	89
FTL	134790	144	100	100	100
FUS	137070	77	100	100	97
FXN	606829	41	100	91	64
GALC	606890	54	100	99	89
GAN	605379	86	100	100	99
GBA	606463	172	100	100	100
GBA2	609471	137	100	100	100
GCDH	608801	112	100	100	100
GCH1	600225	52	100	100	93
GFAP	137780	81	100	100	99
GIGYF2	612003	66	100	99	94
GJC2	608803	85	96	86	76
GLB1	611458	105	100	100	98
GNAL	139312	71	100	100	94
GOSR2	604027	66	100	95	90
GPR143	300808	43	100	89	76
GRID2	602368	78	100	100	99
GRIN1	138249	132	100	100	99
GRIN2B	138252	122	100	100	99
GRM1	604473	136	100	100	99
HACE1	610876	80	100	100	94
HEXB	606873	119	100	100	97
HPCA	142622	248	100	100	100
HPRT1	308000	43	100	99	81
HSD17B4	601860	62	100	98	93
HSPD1	118190	65	100	99	85
HTRA2	606441	143	100	100	98

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IBA57	615316	124	100	100	97
IFRD1	603502	68	100	100	95
ISCA2	615317	89	100	100	100
ITPR1	147265	90	100	100	98
KATNB1	602703	141	100	100	100
KCNA1	176260	126	100	100	100
KCNA2	176262	108	100	100	100
KCNC1	176258	138	100	100	99
KCNC3	176264	91	94	78	64
KCND3	605411	167	100	100	99
KCNJ10	602208	171	100	100	100
KCNMA1	600150	88	100	100	96
KCTD17	616386	89	100	100	97
KCTD7	611725	159	100	100	100
KIDINS220	615759	59	100	98	90
KIF1A	601255	101	100	100	98
KIF1C	603060	141	100	100	100
KIF5A	602821	90	100	100	96
KMT2B	606834	141	97	95	94
L1CAM	308840	101	100	100	100
LAMA1	150320	87	100	100	97
LAMB1	150240	94	100	100	97
LMNB1	150340	66	100	100	97
LRP10	609921	148	100	100	100
LRPPRC	607544	60	100	99	91
LRRK2	609007	78	100	100	96
MAG	159460	156	100	100	100
MAPT	157140	120	100	100	100
MARS2	609728	157	100	100	100
MECP2	300005	113	100	100	96
MECR	608205	97	100	100	99
MED20	612915	148	100	100	100
MICU1	605084	52	100	98	82
MLC1	605908	79	100	100	94
MMADHC	611935	63	100	100	90
MME	120520	58	100	99	91

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MRE11	600814	55	100	97	83
MTHFR	607093	99	100	100	99
MTPAP	613669	97	100	100	97
MTTP	157147	67	100	100	97
MVK	251170	100	100	100	100
MYBPC1	160794	62	100	100	94
MYORG	618255	205	100	100	99
NEFL	162280	157	100	100	100
NEU1	608272	155	100	100	100
NEXMIF	300524	48	100	100	95
NFASC	609145	101	100	100	99
NIPA1	608145	107	100	100	97
NKX2-1	600635	95	100	100	100
NKX6-2	605955	107	99	88	83
NOL3	605235	132	100	100	100
NPC1	607623	87	100	100	99
NPC2	601015	95	100	100	100
NT5C2	600417	54	100	98	89
NUP62	605815	107	100	100	100
OCLN	602876	61	96	84	79
OPA1	605290	59	100	98	89
OPHN1	300127	52	100	96	84
PANK2	606157	81	100	100	99
PARK7	602533	68	100	99	87
PAX6	607108	73	100	100	96
PCNA	176740	93	100	100	92
PDE10A	610652	62	100	99	94
PDE8B	603390	72	100	99	96
PDGFB	190040	101	100	100	100
PDGFRB	173410	116	100	100	100
PDHA1	300502	49	100	94	84
PDHX	608769	79	100	100	97
PDSS1	607429	60	100	93	85
PDSS2	610564	75	100	97	87
PDYN	131340	134	100	100	100
PEX10	602859	100	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PEX2	170993	63	100	100	100
PEX7	601757	57	100	100	95
PHYH	602026	121	100	100	94
PIK3R5	611317	107	100	100	100
PINK1	608309	100	98	93	88
PLA2G6	603604	118	100	100	99
PLD3	615698	148	100	100	100
PLP1	300401	93	100	99	97
PMM2	601785	72	100	100	97
PMPCA	613036	133	100	100	100
PNKD	609023	112	100	100	100
PNKP	605610	104	100	100	98
PNPLA6	603197	123	100	100	99
POLG	174763	114	100	100	99
POLR1C	610060	92	100	100	98
POLR3A	614258	84	100	100	98
POLR3B	614366	76	100	99	93
PPP2R2B	604325	77	100	99	95
PRF1	170280	120	100	100	100
PRICKLE1	608500	76	100	100	98
PRKCG	176980	122	100	100	99
PRKN	602544	96	100	100	99
PRKRA	603424	81	100	100	96
PRRT2	614386	123	100	100	100
PSEN1	104311	59	100	100	93
PTRHD1	617342	119	100	100	100
PUM1	607204	67	100	100	98
PYCR2	616406	114	100	100	100
RAB18	602207	85	100	100	97
RAB29	603949	68	100	100	99
RAB3GAP1	602536	67	100	100	97
RAB3GAP2	609275	66	100	99	92
RAD51	179617	52	88	88	85
RARS1	107820	72	100	98	90
RARS2	611524	66	100	99	93
REEP1	609139	72	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RETREG1	613114	73	100	98	86
RNASEH2A	606034	109	100	100	100
RNASEH2B	610326	62	100	98	88
RNASEH2C	610330	323	100	100	100
RNF170	614649	69	100	97	85
RNF216	609948	66	100	97	86
ROBO3	608630	106	100	100	100
RTN2	603183	135	100	100	100
RUBCN	613516	85	100	98	96
SACS	604490	70	100	100	99
SAMD9L	611170	63	100	100	99
SAMHD1	606754	64	100	99	88
SCN11A	604385	88	100	100	97
SCN8A	600702	110	100	100	98
SCYL1	607982	110	100	100	98
SEPSECS	613009	78	100	100	96
SERAC1	614725	65	100	99	86
SETX	608465	70	100	100	97
SGCE	604149	84	100	100	94
SIL1	608005	100	100	100	100
SLC12A6	604878	67	100	99	94
SLC16A2	300095	75	100	100	95
SLC17A5	604322	86	100	100	97
SLC19A3	606152	73	100	100	98
SLC1A3	600111	99	100	100	100
SLC20A2	158378	99	100	100	95
SLC25A15	603861	132	100	100	98
SLC2A1	138140	127	100	100	100
SLC30A10	611146	148	100	100	100
SLC33A1	603690	68	100	98	86
SLC39A14	608736	92	100	100	97
SLC52A2	607882	173	100	100	100
SLC52A3	613350	109	100	100	100
SLC6A19	608893	115	100	100	100
SLC6A3	126455	102	100	100	100
SLC9A1	107310	119	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC9A6	300231	64	100	96	84
SMPD1	607608	131	100	100	99
SNCA	163890	52	100	100	98
SNORD118	616663	No coverage data			
SNX14	616105	64	100	99	91
SOX10	602229	72	100	97	89
SPART	607111	71	100	100	98
SPAST	604277	64	100	100	94
SPG11	610844	84	100	100	97
SPG21	608181	66	100	95	88
SPG7	602783	125	100	100	97
SPR	182125	102	100	100	100
SPTBN2	604985	121	100	100	100
STUB1	607207	137	100	100	100
SUCLA2	603921	56	100	97	86
SUCLG1	611224	88	100	100	100
SUMF1	607939	88	100	100	99
SUOX	606887	136	100	100	100
SURF1	185620	88	94	89	87
SYNE1	608441	77	100	100	97
SYNJ1	604297	62	100	99	92
SYT14	610949	61	100	95	86
TAF1	313650	54	100	97	87
TANGO2	616830	96	100	100	99
TBC1D20	611663	74	100	93	93
TBC1D23	617687	57	100	99	90
TBCD	604649	104	100	99	96
TBP	600075	80	100	100	96
TDP1	607198	53	100	100	92
TDP2	605764	120	100	100	99
TECPR2	615000	102	100	100	99
TENM4	610084	97	100	99	97
TFG	602498	87	100	100	94
TGM6	613900	117	100	100	99
TH	191290	93	100	99	94
THAP1	609520	87	100	100	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
TIMM8A	300356	138	100	100	100
TMEM106B	613413	72	100	99	89
TMEM230	617019	30	74	52	36
TMEM240	616101	108	100	100	100
TMEM67	609884	77	100	100	94
TOE1	613931	110	100	100	98
TOR1A	605204	73	100	100	98
TPP1	607998	97	100	100	100
TREM2	605086	107	100	100	100
TREX1	606609	234	100	100	100
TRPC3	602345	112	100	99	95
TSEN2	608753	63	87	86	78
TSEN54	608755	105	100	96	96
TTBK2	611695	74	100	100	99
TTC19	613814	53	100	92	77
TTPA	600415	61	100	98	91
TUBA1A	602529	110	100	100	100
TUBB4A	602662	242	100	100	99
TUBG1	191135	190	100	100	100
TWNK	606075	145	100	100	100
TYROBP	604142	108	100	100	100
UBA5	610552	46	100	91	67
UBAP1	609787	73	98	92	86
UBQLN2	300264	79	100	100	100
UCHL1	191342	79	100	100	99
VAMP1	185880	104	100	100	100
VCP	601023	90	100	100	98
VLDLR	192977	70	100	100	97
VPS13A	605978	72	100	99	91
VPS13C	608879	50	100	97	85
VPS13D	608877	64	100	98	92
VPS35	601501	68	100	99	94
VPS37A	609927	46	100	99	86
VPS53	615850	82	100	100	96
VRK1	602168	53	100	99	92
VWA3B	614884	66	100	98	92

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
WASHC5	610657	62	100	100	94
WDR26	617424	50	100	94	81
WDR45	300526	92	100	100	100
WDR73	616144	152	100	100	97
WDR81	614218	155	100	100	100
WFS1	606201	196	100	100	100
WWOX	605131	92	100	100	100
XK	314850	62	100	100	96
XPR1	605237	64	100	100	99
XRCC1	194360	97	100	100	100
ZC4H2	300897	78	100	99	93
ZFYVE26	612012	90	100	100	98
ZFYVE27	610243	99	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