

Whole Exome Sequencing

Gene package Movement disorders, version 9, 26-2-2021



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/>). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: <http://www.broadinstitute.org/gatk/>). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). 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 addition to either MPLA and/or exome sequencing. It is not excluded that pathogenic variants 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
AAAS	605378	100	100	100	99.57
AARS1	601065	100	100	100	98.97
AARS2	612035	100	99.76	98.92	92.46
ABCA2	600047	100	100	99.42	94.86
ABCB7	300135	100	100	98.76	90.71
ABCD1	300371	100	96.08	92.22	83.26
ABHD12	613599	99.76	95.79	90.25	80.60
ACTB	102630	100	100	100	98.33
ADAR	146920	98.45	98.45	98.45	97.45
ADCY5	600293	99.35	97.75	95.86	88.53
ADGRG1	604110	100	98.85	95.55	81.55
ADPRHL2	610624	100	100	98.92	91.94
AFG3L2	604581	95.13	91.50	91.50	89.77
AGTPBP1	606830	97.70	94.22	91.44	85.90
AIMP1	603605	100	100	94.51	77.98
ALDH18A1	138250	100	100	99.65	96.64

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ALDH3A2	609523	100	100	99.01	94.61
ALDH5A1	610045	100	99.09	92.14	79.75
ALS2	606352	100	99.96	98.79	93.76
ANO10	613726	100	100	99.96	94.91
ANO3	610110	100	100	99.81	95.32
AP4B1	607245	100	100	100	98.97
AP4E1	607244	100	100	99.77	95.61
AP4M1	602296	100	99.70	95.60	79.02
AP4S1	607243	100	100	100	96.10
AP5Z1	613653	100	99.04	94.73	76.89
APTX	606350	100	100	99.44	93.80
ARG1	608313	100	100	100	96.51
ARL6IP1	607669	98.49	80.80	80.80	80.80
ARSA	607574	100	100	100	100
ARX	300382	85.61	80.04	71.89	58.82
ASPA	608034	100	100	100	100
ATCAY	608179	100	100	100	98.52
ATG5	604261	100	100	100	97.10
ATL1	606439	100	100	100	98.90
ATM	607585	100	94.70	74.49	49.62
ATN1	607462	100	97.92	94.71	81.97
ATP13A2	610513	100	99.76	96.37	77.46
ATP1A2	182340	100	100	99.46	93.95
ATP1A3	182350	100	100	99.04	92.02
ATP2B3	300014	100	99.70	98.41	92.60
ATP6AP2	300556	100	100	96.34	81.63
ATP7B	606882	100	100	98.95	94.62
ATP8A2	605870	100	100	99.18	94.45
ATXN1	601556	100	100	100	100
ATXN2	601517	95.40	89.43	85.04	77.05
ATXN3	607047	100	100	97.67	93.26
ATXN7	607640	96.93	95.06	93.48	89.07
AUH	600529	100	100	100	100
B4GALNT1	601873	100	97.19	94.52	86.71
BCAP31	300398	100	100	97.49	82.40
BCKDHA	608348	100	98.91	95.61	88.71
BCKDHB	248611	100	100	100	99.56
BCS1L	603647	100	100	100	100
BSCL2	606158	100	99.47	97.07	90.19
C12orf65	613541	100	100	92.57	56.13

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C19orf12	614297	100	100	100	100
C9orf72	614260	100	100	100	99.33
CA8	114815	100	100	100	96.28
CACNA1A	601011	99.96	98.08	92.14	74.91
CACNA1B	601012	97.03	95.07	92.44	86.46
CACNA1E	601013	100	99.94	99.54	97.17
CACNA1G	604065	100	99.47	97.97	91.69
CACNB4	601949	100	95.49	95.08	90.62
CAMTA1	611501	98.81	98.61	96.50	90.59
CAPN1	114220	100	99.38	95.78	88.39
CCDC88C	611204	100	99.87	98.51	91.23
CCT5	610150	100	100	100	98.70
CDKL5	300203	100	97.89	94.24	84.71
CHCHD2	616244	100	100	98.63	86.64
CHMP1A	164010	100	100	98.93	79.05
CHP1	606988	100	100	100	98.76
CHRNA4	118504	100	95.20	95.20	94.53
CHRN2	118507	100	100	98.37	93.82
CIZ1	611420	100	94.37	85.61	69.83
CLCN2	600570	100	100	99.73	94.38
CLCN4	302910	100	100	98.98	90.40
CLP1	608757	100	100	100	100
CLPB	616254	100	100	100	98.07
COASY	609855	100	100	98.18	91.86
COL6A3	120250	100	100	99.43	98.34
COQ2	609825	100	99.47	93.83	74.02
COQ8A	606980	100	100	100	95.72
COQ9	612837	100	96.96	91.80	82.63
COX10	602125	100	100	99.59	96.26
COX15	603646	100	100	97.36	90.10
COX20	614698	100	95.71	85	56.12
CP	117700	99.05	99.05	99.05	98.28
CPT1C	608846	100	99.08	95.11	78.02
CSF1R	164770	100	99.64	97.42	87.66
CSTB	601145	100	100	99.58	75.71
CWF19L1	616120	100	100	99.58	92.87
CYB5R3	613213	100	99.70	95.22	79.43
CYP27A1	606530	100	100	100	100
CYP2U1	610670	99.25	95.64	92.96	87.82
CYP7B1	603711	100	98.99	92.55	91.30

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
DAB1	603448	100	100	100	100
DBT	248610	100	99.94	98.65	92.80
DCAF17	612515	100	100	98.51	91.39
DCC	120470	100	100	99.58	95.80
DCTN1	601143	100	99.45	96.67	91.43
DDC	107930	100	98.76	92.47	78.85
DDHD1	614603	99.81	97.85	95.57	92.94
DDHD2	615003	100	100	100	95.53
DHDDS	608172	100	100	99.17	91.25
DLAT	608770	100	99.55	97.95	92.41
DLD	238331	100	100	99.97	94.83
DNAJC13	614334	100	99.96	99.47	96.31
DNAJC3	601184	100	100	98.92	89.47
DNAJC6	608375	100	100	99.13	93.47
DNAL4	610565	100	100	100	91.47
DNMT1	126375	100	99.55	97.62	91.57
DSTYK	612666	100	100	99.51	96.36
ECHS1	602292	100	100	94.03	89.51
EEF2	130610	100	100	99.77	95.08
EIF2AK2	176871	100	100	99.08	93.29
EIF2B1	606686	100	100	99.82	94.75
EIF2B2	606454	100	99.75	98.43	94.15
EIF2B3	606273	100	100	100	99.40
EIF2B4	606687	100	98.64	93.94	90.73
EIF2B5	603945	100	98.45	96.64	91.14
EIF4G1	600495	100	99.24	98.31	92.36
ELOVL4	605512	100	100	98.49	92.33
ELOVL5	611805	100	100	100	99.45
ENTPD1	601752	100	100	100	98.47
ERLIN1	611604	100	100	100	100
ERLIN2	611605	100	100	100	100
EXOSC3	606489	100	100	100	99.23
FA2H	611026	100	94.15	83.56	67.28
FAM126A	610531	100	100	100	99.66
FAR1	616107	100	100	100	98.30
FARS2	611592	100	100	100	97.59
FAT2	604269	100	99.88	99.69	98.42
FBXO7	605648	100	94.44	91.87	90.41
FGF14	601515	100	100	100	99.42
FLVCR1	609144	100	98.87	95.36	86.43

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FMR1	309550	100	98.03	92.62	72.16
FOLR1	136430	100	100	100	98.88
FOXG1	164874	98.76	94.08	91.12	86.79
FRMD7	300628	100	100	100	97.82
FTL	134790	100	100	95.54	85.54
FUS	137070	100	100	99.33	92.92
FXN	606829	100	99.04	94.93	76.71
GALC	606890	99.96	99.96	99.54	92.36
GAN	605379	100	99.95	98.76	95.67
GBA	606463	100	100	100	98.63
GBA2	609471	100	100	98.99	97.18
GCDH	608801	100	100	99.22	93.55
GCH1	600225	94.02	94.02	94.02	91.15
GFAP	137780	100	99.60	95.17	81.90
GIGYF2	612003	100	97.38	91.42	82.23
GJC2	608803	98.92	91.29	81.49	61.37
GLB1	611458	100	100	99.78	97.42
GNAL	139312	100	100	100	100
GOSR2	604027	100	100	99.95	94.46
GPR143	300808	99.64	84.55	80.60	75.68
GRID2	602368	100	100	99.61	97.96
GRIN1	138249	100	100	97.17	90.83
GRIN2B	138252	100	100	99.87	98.02
GRM1	604473	100	100	99.68	98.01
HACE1	610876	100	99.53	96.07	90.12
HEXB	606873	99.00	95.92	92.79	85.27
HPCA	142622	100	100	100	95.23
HPRT1	308000	95.44	92.33	85.67	71.16
HSD17B4	601860	98.96	97.05	94.86	87.75
HSPD1	118190	100	100	98.50	91.23
HTRA2	606441	100	100	100	98.96
IBA57	615316	100	100	100	100
IFRD1	603502	100	99.44	97.77	83.96
IRF2BPL	611720	98.53	96.22	94.39	89.99
ISCA2	615317	100	100	100	100
ITPR1	147265	100	99.72	98.63	94.71
JAM2	606870	100	91.10	90.99	85.82
KATNB1	602703	100	100	100	98.02
KCNA1	176260	100	100	100	97.51
KCNA2	176262	100	100	100	100

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KCNC1	176258	100	100	100	100
KCNC3	176264	86.07	78.88	72.31	59.19
KCND3	605411	100	100	100	96.34
KCNJ10	602208	100	100	100	100
KCNMA1	600150	100	100	93.27	71.95
KCTD17	616386	98.99	91.38	80.44	55.66
KCTD7	611725	100	100	100	98.20
KIDINS220	615759	100	99.31	97.52	92.89
KIF1A	601255	100	100	99.61	91.46
KIF1C	603060	100	99.50	97.43	90.90
KIF5A	602821	100	100	98.81	90.10
KMT2B	606834	95.68	94.87	92.98	86.63
L1CAM	308840	100	100	99.58	93.95
LAMA1	150320	100	100	99.64	96.22
LAMB1	150240	100	100	99.63	97.11
LMNB1	150340	100	99.04	95.55	87.41
LRP10	609921	100	99.41	96.88	89.12
LRPPRC	607544	99.94	98.73	97.72	93.88
LRRK2	609007	99.83	99.28	98.31	94.84
MAG	159460	100	99.64	97.50	89.50
MAPT	157140	100	99.35	96.51	84.22
MARS2	609728	100	100	100	100
MECP2	300005	100	99.68	97.39	91.06
MECR	608205	99.10	93.11	86.15	80.99
MED20	612915	100	100	100	98.60
MICU1	605084	100	100	100	97.95
MLC1	605908	100	100	99.07	85.27
MMADHC	611935	100	100	99.90	88.42
MME	120520	100	81.33	76.50	56
MRE11	600814	100	100	99.52	95.09
MTHFR	607093	100	100	99.06	94.14
MTPAP	613669	100	98.83	95.12	81.75
MTPP	157147	100	100	100	99.06
MVK	251170	100	100	100	96.47
MYBPC1	160794	100	100	100	96.53
MYORG	618255	100	100	100	99.03
NDUFA10	603835	94.60	92.83	92.08	88.53
NDUFA12	614530	100	100	100	94.47
NDUFA2	602137	100	100	100	100
NDUFA9	603834	100	100	100	100

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NDUFS1	157655	100	100	100	98.48
NDUFS2	602985	100	100	99.58	91.61
NDUFS3	603846	100	100	100	100
NDUFS4	602694	100	100	100	100
NDUFS6	603848	100	100	96.02	83.30
NDUFS7	601825	100	98.62	91.93	66.40
NDUFS8	602141	100	98.80	95	85.80
NEFL	162280	99.88	99.48	96.65	88.09
NEU1	608272	100	100	100	99.05
NEXMIF	300524	100	100	100	99.31
NFASC	609145	100	99.63	97.72	91.50
NIPA1	608145	100	100	99.70	94.12
NKX2-1	600635	100	100	97.55	73.71
NKX6-2	605955	100	93.49	85.75	73.63
NOL3	605235	100	100	100	91.29
NOTCH2NLC	618025	No coverage data			
NPC1	607623	100	99.90	98.22	94.90
NPC2	601015	100	100	100	99.42
NT5C2	600417	99.59	97.87	95.86	86.62
NUP62	605815	100	100	100	90.23
OCLN	602876	72.78	71.22	63.56	47.48
OPA1	605290	100	99.07	96.09	86.31
OPHN1	300127	100	100	99.53	87.77
PAH	612349	100	100	100	98.21
PANK2	606157	100	100	100	94.93
PARK7	602533	100	100	100	85.52
PAX6	607108	100	100	100	99.85
PCBD1	126090	100	98.21	94.13	94.13
PCCB	232050	100	100	99.17	92.49
PCNA	176740	100	100	100	98.73
PCYT2	602679	100	92.90	92.44	87.64
PDE10A	610652	100	99.86	97.03	90.58
PDE8B	603390	100	100	98.73	89.35
PDGFB	190040	100	100	100	89.92
PDGFRB	173410	100	100	99.25	93.49
PDHA1	300502	100	98.69	94.91	80.12
PDHB	179060	100	100	100	95.81
PDHX	608769	100	99.89	93.21	80.89
PDSS1	607429	100	100	100	99.77
PDSS2	610564	100	99.04	97.05	92.74

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PDYN	131340	100	100	97.76	91.15
PEX10	602859	100	100	96.80	80.48
PEX2	170993	100	100	100	100
PEX7	601757	100	100	96.83	87.17
PHYH	602026	99.89	89.33	89.33	76.91
PIK3R5	611317	100	100	98.10	87.47
PINK1	608309	98.32	93.93	88.81	75.72
PLA2G6	603604	100	100	99.38	92.46
PLD3	615698	100	99.11	96.24	88.85
PLP1	300401	100	99.59	95.57	85.43
PMM2	601785	100	100	100	95.16
PMPCA	613036	100	100	98.01	91.28
PNKD	609023	100	100	100	95.99
PNKP	605610	100	99.95	98.30	91.85
PNPLA6	603197	100	98.93	95.55	88.18
POLG	174763	100	99.94	97.83	89.26
POLR1C	610060	100	100	100	92.16
POLR3A	614258	100	100	99.87	98.36
POLR3B	614366	100	100	99.10	97.16
PPP2R2B	604325	100	99.57	90.80	86.52
PRF1	170280	100	100	100	100
PRICKLE1	608500	100	100	99.66	97.30
PRKCG	176980	100	100	99.40	96.59
PRKN	602544	100	99.20	97.25	91.68
PRKRA	603424	100	100	100	100
PRRT2	614386	100	100	100	97.38
PSEN1	104311	100	100	100	96.28
PTRHD1	617342	100	100	100	100
PTS	612719	100	100	100	100
PUM1	607204	100	100	99.65	97.69
PYCR2	616406	100	100	96.85	77.33
QDPR	612676	100	100	99.77	87.56
RAB18	602207	97.51	89.83	86.24	75.72
RAB29	603949	100	100	100	98.03
RAB3GAP1	602536	100	100	100	97.96
RAB3GAP2	609275	100	98.46	98.05	94.92
RAD51	179617	88.25	88.25	88.25	88.25
RARS1	107820	100	99.96	99.39	93.51
RARS2	611524	100	99.98	99.18	96.73
REEP1	609139	100	100	97.71	83.57

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RETREG1	613114	99.07	95.12	92.28	87.52
RNASEH2A	606034	100	100	98.86	85.19
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	97.20
RNF170	614649	100	100	99.33	91.00
RNF216	609948	100	100	99.84	97.23
ROBO3	608630	100	99.62	97.14	82.24
RTN2	603183	100	100	100	99.21
RUBCN	613516	100	99.90	97.88	90.43
SACS	604490	100	100	100	99.92
SAMD9L	611170	100	100	100	100
SAMHD1	606754	100	100	100	99.98
SCN11A	604385	100	99.99	99.68	97.85
SCN2A	182390	100	99.96	99.36	94.90
SCN8A	600702	100	100	99.53	95.79
SCO2	604272	100	100	100	100
SCP2	184755	100	100	100	99.43
SCYL1	607982	100	100	98.65	85.60
SEMA6B	608873	97.15	87.03	78.75	68.83
SEPSECS	613009	100	100	100	98.64
SERAC1	614725	100	100	100	97.87
SETX	608465	100	100	100	99.08
SGCE	604149	100	100	100	96.71
SIL1	608005	100	99.49	96.74	87.91
SLC12A6	604878	100	99.51	98.56	93.33
SLC16A2	300095	100	99.86	93.26	86.50
SLC17A5	604322	100	100	100	95.37
SLC19A3	606152	99.94	99.94	99.94	98.87
SLC1A3	600111	100	100	100	80.93
SLC20A2	158378	100	100	97.56	93.16
SLC25A15	603861	100	100	100	100
SLC2A1	138140	100	97.73	97.73	97.43
SLC30A10	611146	100	100	100	97.98
SLC33A1	603690	100	100	99.77	98.02
SLC39A14	608736	97.92	97.92	96.77	89.26
SLC52A2	607882	100	100	100	95.83
SLC52A3	613350	100	100	99.39	95.90
SLC6A19	608893	100	98.09	91.36	81.26
SLC6A3	126455	100	99.86	98.27	89.02
SLC9A1	107310	100	99.01	97.19	90.13

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SLC9A6	300231	100	98.43	96.80	87.64
SMPD1	607608	100	100	100	97.86
SNCA	163890	100	100	100	98.85
SNORD118	616663	No coverage data			
SNX14	616105	99.60	93.87	86.76	68.39
SOX10	602229	100	98.83	96.64	87.59
SPART	607111	100	100	99.86	96.80
SPAST	604277	100	98.99	94.56	80.16
SPG11	610844	100	99.96	99.19	95.17
SPG21	608181	100	100	97.30	84.64
SPG7	602783	91.82	90.83	88.09	82.63
SPR	182125	100	100	95.49	79.89
SPTBN2	604985	99.97	98.14	94.29	85.18
STUB1	607207	100	100	100	100
SUCLA2	603921	100	100	99.94	98.79
SUCLG1	611224	100	100	100	94.58
SUMF1	607939	100	100	100	100
SUOX	606887	100	100	100	96.73
SURF1	185620	100	100	100	96.29
SYNE1	608441	100	99.63	98.33	91.99
SYNJ1	604297	99.02	95.04	94.09	90.75
SYT14	610949	100	99.08	97.23	93.15
TACO1	612958	100	100	96.17	83.45
TAF1	313650	100	99.87	98.87	93.03
TANGO2	616830	100	100	99.90	96.00
TBC1D20	611663	98.94	93.43	93.43	92.99
TBC1D23	617687	100	99.92	98.71	92.01
TBCD	604649	98.59	95.32	92.79	83.69
TBP	600075	100	100	100	98.70
TDP1	607198	100	100	99.81	96.19
TDP2	605764	100	100	100	93.88
TECPR2	615000	100	100	99.62	95.92
TENM4	610084	100	99.48	97.56	92.66
TFG	602498	100	100	99.81	92.69
TGM6	613900	100	100	99.54	94.20
TH	191290	100	97.40	93.00	83.58
THAP1	609520	100	100	100	99.28
THG1L	618802	100	100	100	99.58
TIMM8A	300356	100	100	100	92.75
TMEM106B	613413	100	100	100	91.32

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
TMEM230	617019	80.59	80.59	80.05	71.03
TMEM240	616101	100	100	97.58	87.15
TMEM67	609884	100	100	99.55	88.82
TOE1	613931	100	100	100	100
TOR1A	605204	100	100	100	99.27
TPP1	607998	100	99.44	97.38	93.18
TRAPPC4	610971	100	100	100	100
TREM2	605086	100	100	100	97.22
TREX1	606609	100	100	100	100
TRPC3	602345	99.83	97.75	95.52	90.81
TSEN2	608753	86.99	86.99	86.99	85.72
TSEN54	608755	95.91	95.77	93.60	83.12
TTBK2	611695	100	100	100	99.21
TTC19	613814	100	100	100	95.05
TTPA	600415	99.89	97.70	91.43	75.59
TUBA1A	602529	100	100	100	100
TUBB4A	602662	100	94.57	87.57	87.57
TUBG1	191135	100	100	100	100
TWINK	606075	100	100	100	100
TYROBP	604142	100	100	88.50	69.48
UBA5	610552	97.07	90.98	87.53	73.90
UBAP1	609787	99.59	96.18	91.14	82.10
UBQLN2	300264	100	100	100	99.10
UCHL1	191342	100	89.10	82.45	74.20
UGDH	603370	100	100	99.94	94.83
VAMP1	185880	100	100	92.51	92.51
VAMP2	185881	100	100	92.51	90.20
VCP	601023	100	100	99.69	98.66
VLDLR	192977	100	99.97	97.85	95.18
VPS13A	605978	100	98.91	97.29	88.56
VPS13C	608879	98.39	97.54	96.23	91.69
VPS13D	608877	100	100	99.63	96.24
VPS35	601501	100	100	99.71	97.78
VPS37A	609927	100	98.94	96.60	88.24
VPS53	615850	100	100	100	96.29
VRK1	602168	100	100	100	93.10
VWA3B	614884	100	99.56	96.74	93.91
WASHC5	610657	100	100	100	96.15
WDR26	617424	100	97.41	95.76	90.46
WDR45	300526	100	100	100	98.17

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
WDR73	616144	100	100	100	98.57
WDR81	614218	100	99.31	97.09	88.73
WFS1	606201	100	100	99.40	96.23
WWOX	605131	100	100	97.53	77.29
XK	314850	100	100	100	97.81
XPR1	605237	100	100	99.62	95.81
XRCC1	194360	100	98.75	94.15	80.50
ZC4H2	300897	100	100	100	96.37
ZFYVE26	612012	100	99.70	98.43	93.21
ZFYVE27	610243	100	100	97.24	88.41

- OMIM release used: 18-2-2021

- The statistics above are based on a set of 100 samples

- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence ($\pm 10bp$ flanking introns) that is covered at least 10x, 20x, 30x or 50x