

# Whole Exome Sequencing

## Gene package Neuronal migration 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 several (fragments of) genes involved in neuronal migration disorders (SALSA P061 Lissencephaly; MRC Holland). 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
ACTB	102630	100	100	100	98.33
ACTG1	102560	100	100	100	100
ADA2	607575	100	100	100	99.95
ADAR	146920	98.45	98.45	98.45	97.45
ADGRG1	604110	100	98.85	95.55	81.55
AGBL2	617345	98.87	95.83	94.37	87.67
AGTPBP1	606830	97.70	94.22	91.44	85.90
AKT1	164730	100	100	100	93.53
AKT3	611223	100	100	100	95.64
ANKLE2	616062	98.91	96.19	92.18	88.31
AP1S2	300629	100	94.91	87.33	60.78
AP3B2	602166	100	98.57	83.56	74.57
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
APC2	612034	98.39	94.14	88.60	75.10
ARFGEF2	605371	100	100	99.45	95.27
ARNT2	606036	100	100	99.41	95.22
ARX	300382	85.61	80.04	71.89	58.82
ASNS	108370	100	100	99.47	94.75
ASPM	605481	99.82	99.07	97.76	92.66
ASXL1	612990	98.42	98.42	98.03	95.44
ATAD3A	612316	95.36	88.37	85.31	81.23

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ATAD3B	612317	94.94	88.56	85.58	82.20
ATP1A2	182340	100	100	99.46	93.95
ATP1A3	182350	100	100	99.04	92.02
ATP6VOA2	611716	100	99.66	98.26	92.39
ATR	601215	100	99.95	99.23	94.76
ATRIP	606605	100	100	99.41	94.00
B3GALNT2	610194	100	100	99.57	92.82
B4GAT1	605517	100	100	100	100
BAP1	603089	100	99.98	97.63	91.41
CASK	300172	100	97.33	93.54	76.84
CCND2	123833	100	98.97	94.67	80.82
CDK5	123831	100	100	100	93.23
CDK5RAP2	608201	99.95	98.96	96.74	91.89
CDK6	603368	100	100	99.60	91.73
CENPJ	609279	100	100	100	99.23
CEP135	611423	100	98.78	93.77	75.53
CEP152	613529	97.93	97.58	95.74	90.55
CEP63	614724	100	98.70	92.80	75.24
CHMP1A	164010	100	100	98.93	79.05
CIT	605629	100	99.59	98.47	94.80
CLEC16A	611303	99.95	98.15	95.22	88.11
CLP1	608757	100	100	100	100
COL18A1	120328	100	99.77	98.50	85.10
COL4A1	120130	99.06	97.00	93.11	83.56
COL4A2	120090	100	99.28	97.30	88.89
COLGALT1	617531	96.23	84.78	80.65	69.06
CRADD	603454	100	100	97.10	89.32
CRB2	609720	100	99.09	97.41	85.09
CRPPA	614631	100	97.30	94.46	82.16
CSTB	601145	100	100	99.58	75.71
CTC1	613129	100	100	99.31	90.43
CTNNA2	114025	100	99.50	97.64	89.24
CTNND2	604275	94.08	91.56	88.47	81.03
DAB1	603448	100	100	100	100
DAG1	128239	100	100	98.72	96.62
DCHS1	603057	100	100	100	98.84
DCX	300121	100	98.56	95.83	82.33
DDX3X	300160	100	100	99.27	88.84
DEPDC5	614191	100	99.84	98.86	95.68
DKC1	300126	100	98.05	94.16	83.85
DNMT3A	602769	100	99.28	90.86	70.43
DYNC1H1	600112	100	99.77	98.94	95.30
EIF2AK3	604032	99.92	97.00	93.56	86.97
EMG1	611531	100	100	100	98.96
EML1	602033	100	97.06	97.06	95.09

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EOMES	604615	100	100	93.71	83.11
ERCC1	126380	100	98.98	91.42	54.24
ERCC2	126340	100	100	99.69	90.71
ERCC5	133530	100	98.96	95.87	87.36
ERCC6	609413	100	99.55	98.93	96.63
ERMARD	615532	100	98.91	97.39	91.08
FAT4	612411	100	99.99	99.92	99.24
FIG4	609390	100	100	100	98.04
FKRP	606596	100	100	100	97.31
FKTN	607440	100	100	99.39	91.81
FLNA	300017	100	99.43	98.63	95.60
FLVCR2	610865	100	100	100	99.21
FOXC1	601090	95.68	89.67	84.81	72.75
FRMD4A	616305	100	99.61	98.19	92.15
G3BP1	608431	100	98.05	94.53	77.26
G3BP2	No ID	100	100	100	97.72
GNAQ	600998	100	100	100	96.34
HNRNPK	600712	100	99.82	96.69	86.20
IBA57	615316	100	100	100	100
IER3IP1	609382	100	100	100	94.28
IFIH1	606951	100	100	100	97.67
INTS8	611351	99.52	96.98	92.72	85.42
ITSN1	602442	100	99.35	96.68	89.32
JAM3	606871	100	100	100	97.23
KATNB1	602703	100	100	100	98.02
KIF11	148760	100	98.12	94.21	84.38
KIF13B	607350	100	99.90	98.79	93.69
KIF2A	602591	100	99.68	95.44	84.80
KIF5C	604593	99.73	96.86	91.87	82.45
KIF7	611254	99.65	96.04	90.61	78.96
KIFBP	609367	100	100	100	88.42
KNL1	609173	99.28	99.28	98.62	97.57
KPTN	615620	100	100	97.06	83.17
L1CAM	308840	100	100	99.58	93.95
LAMA1	150320	100	100	99.64	96.22
LAMA2	156225	100	99.94	99.12	93.86
LAMB1	150240	100	100	99.63	97.11
LAMC1	150290	100	98.97	97.38	92.15
LAMC3	604349	100	99.07	96.44	83.66
LARGE1	603590	100	100	98.94	95.21
LARP7	612026	100	96.99	90.87	73.88
MACF1	608271	100	99.83	98.96	94.20
MAP1A	600178	100	100	98.97	91.76
MCF2L	609499	100	99.82	97.64	88.53
MCPH1	607117	98.36	95.86	91.18	87.40

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MDGA1	609626	100	99.31	96.94	88.16
MED13	603808	100	99.54	98.76	94.55
MED17	603810	100	99.84	98.22	87.98
MN1	156100	100	100	99.94	88.92
MPDZ	603785	100	100	99.71	97.15
MTOR	601231	100	99.73	99.19	94.88
MYCN	164840	100	100	97.62	91.49
NBN	602667	100	100	100	96.80
NCAPD2	615638	100	100	99.56	95.26
NCAPD3	609276	100	100	99.80	96.91
NCAPH	602332	100	97.27	94.31	90.50
NCAPH2	611230	100	100	98.78	91.91
NDE1	609449	100	100	95.71	82.83
NEDD4L	606384	100	99.41	96.38	90.03
NFIA	600727	100	98.58	95.22	87.59
NID1	131390	100	100	98.95	90.57
NIN	608684	100	99.86	98.83	95.18
NPRL2	607072	100	100	100	98.27
NPRL3	600928	100	100	97.66	78.76
NSDHL	300275	100	100	100	89.20
OCLN	602876	72.78	71.22	63.56	47.48
PAFAH1B1	601545	100	95.52	93.36	92.38
PAX6	607108	100	100	100	99.85
PCDH12	605622	100	100	99.92	95.55
PCNT	605925	100	99.35	96.70	90.36
PHC1	602978	100	99.10	92.68	85.01
PHGDH	606879	100	100	99.35	93.12
PI4KA	600286	100	98.18	94.78	88.52
PIK3CA	171834	100	100	99.94	97.86
PIK3R2	603157	93.24	90.46	88.47	84.00
PLK4	605031	100	100	99.26	95.79
PNKP	605610	100	99.95	98.30	91.85
POLR3B	614366	100	100	99.10	97.16
POMGNT1	606822	100	100	100	98.92
POMT1	607423	100	100	98.66	94.11
POMT2	607439	100	100	99.66	93.95
PRUNE1	617413	100	100	100	100
PSAT1	610936	100	100	98.22	89.15
PSPH	172480	100	96.26	90.32	72.97
PTEN	601728	100	100	100	100
PTF1A	607194	100	91.46	78.32	50.63
PYCR2	616406	100	100	96.85	77.33
QARS1	603727	100	100	100	97.54
RAB18	602207	97.51	89.83	86.24	75.72
RAB3GAP1	602536	100	100	100	97.96

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RAB3GAP2	609275	100	98.46	98.05	94.92
RAD50	604040	100	98.40	92.28	77.78
RARS2	611524	100	99.98	99.18	96.73
RBBP8	604124	100	99.77	98.87	92.85
RBM10	300080	100	99.32	96.80	89.28
RELN	600514	100	99.99	99.49	95.45
RHEB	601293	92.42	89.89	89.04	72.40
RNASEH2A	606034	100	100	98.86	85.19
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	97.20
RNASET2	612944	100	98.58	94.83	82.01
RNU4ATAC	601428	No coverage data			
ROBO3	608630	100	99.62	97.14	82.24
RTEL1	608833	100	100	99.24	90.23
RTTN	610436	100	100	99.58	92.62
RXYLT1	605862	100	100	100	100
SAMHD1	606754	100	100	100	99.98
SCN3A	182391	100	100	100	98.05
SHANK3	606230	94.15	88.94	81.69	65.49
SHOC2	602775	100	100	100	97.82
SLC25A19	606521	100	100	99.26	96.25
SLC35A2	314375	100	100	95.84	84.43
SMPD4	610457	100	98.90	96.31	84.36
SNAP29	604202	100	100	100	92.79
SRPX2	300642	100	100	98.81	94.55
STAMBP	606247	100	100	100	98.38
STIL	181590	100	100	100	98.86
STRADA	608626	100	100	100	94.98
TBC1D20	611663	98.94	93.43	93.43	92.99
TBC1D24	613577	100	100	99.15	96.48
TBC1D7	612655	100	100	100	97.82
TBR1	604616	100	100	100	97.69
TMTC3	617218	100	99.25	95.90	85.39
TMX2	616715	100	100	100	100
TRAIP	605958	100	100	100	100
TREX1	606609	100	100	100	100
TSC1	605284	100	100	100	98.90
TSC2	191092	100	100	99.62	95.03
TSEN54	608755	95.91	95.77	93.60	83.12
TUBA1A	602529	100	100	100	100
TUBA8	605742	98.41	98.41	98.41	97.65
TUBB	191130	100	100	100	95.10
TUBB2A	615101	100	90.88	80.64	65.62
TUBB2B	612850	100	93.85	81.98	68.13
TUBB3	602661	100	99.79	94.62	94.62

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TUBB4A	602662	100	94.57	87.57	87.57
TUBG1	191135	100	100	100	100
TUBGCP4	609610	100	100	99.30	94.73
TUBGCP6	610053	100	100	99.87	97.16
VLDLR	192977	100	99.97	97.85	95.18
VPS13B	607817	99.15	98.02	95.90	88.67
VRK1	602168	100	100	100	93.10
WASHC5	610657	100	100	100	96.15
WDR4	605924	100	100	96.09	80.43
WDR62	613583	100	99.64	98.34	91.23
WDR73	616144	100	100	100	98.57
WDR81	614218	100	99.31	97.09	88.73
YWHAE	605066	100	95.54	90.06	82.88
ZIC1	600470	100	100	99.29	95.43
ZIC2	603073	93.72	90.64	88.47	83.88
ZIC4	608948	100	100	100	100

- 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 (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x