

Whole Exome Sequencing

Gene package Neuronal migration disorders, version 10, 30-9-2021



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 $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
ACTB	102630	100	100	100	99.59
ACTG1	102560	100	100	100	100
ADA2	607575	100	100	100	100
ADAR	146920	98.45	98.45	98.45	98.30
ADGRG1	604110	100	99.08	96.19	85.38
AGBL2	617345	99.87	97.83	95.30	88.98
AGTPBP1	606830	98.52	95.12	91.67	86.53
AKT1	164730	100	100	100	98.26
AKT3	611223	100	100	100	97.02
ANKLE2	616062	98.57	94.60	92.49	89.95
AP1S2	300629	100	99.91	91.29	75.69
AP3B2	602166	100	98.95	82.50	74.57
AP4B1	607245	100	100	100	100
AP4E1	607244	100	100	100	97.99
AP4M1	602296	100	100	96.46	81.67
AP4S1	607243	100	100	100	100
APC2	612034	98.41	94.37	91.08	78.13
ARFGEF2	605371	100	100	100	98.69
ARNT2	606036	100	100	98.72	95.99
ARX	300382	83.87	74.50	64.03	41.41

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ASNS	108370	100	100	100	97.53
ASPM	605481	100	99.21	97.94	93.28
ASXL1	612990	98.42	98.42	98.42	97.45
ATAD3A	612316	95.68	89.87	86.81	82.45
ATAD3B	612317	95.41	90.06	87.19	83.50
ATP1A2	182340	100	100	100	97.98
ATP1A3	182350	100	100	99.43	95.12
ATP6V0A2	611716	100	100	99.23	94.78
ATR	601215	100	100	99.66	95.63
ATRIP	606605	100	100	100	98.58
B3GALNT2	610194	100	100	100	92.67
B4GAT1	605517	100	100	100	100
BAP1	603089	100	99.80	98.20	94.42
CASK	300172	100	99.04	96.53	85.71
CBY1	607757	100	100	100	100
CCND2	123833	100	99.69	96.07	84.38
CD40	109535	100	100	100	100
CDK5	123831	100	100	100	94.89
CDK5RAP2	608201	100	99.50	98.31	94.35
CDK6	603368	100	100	100	94.36
CENPJ	609279	100	100	100	99.46
CEP135	611423	100	99.73	96.72	81.09
CEP152	613529	97.93	97.71	95.63	92.34
CEP63	614724	100	100	96.71	84.05
CEP85L	618865	98.08	94.49	90.83	86.10
CHMP1A	164010	100	100	100	88.57
CIT	605629	100	99.79	99.21	96.50
CLEC16A	611303	100	99.15	97.39	92.62
CLP1	608757	100	100	100	100
COL18A1	120328	100	99.83	98.32	86.94
COL4A1	120130	98.87	97.12	94.05	85.47
COL4A2	120090	100	99.92	99.04	92.59
COLGALT1	617531	97.20	84.85	82.41	73.08
CRADD	603454	100	100	99.84	93.56
CRB2	609720	100	99.53	97.95	88.29
CRPPA	614631	100	99.97	97.01	87.99
CSTB	601145	100	100	90.82	75.71
CTC1	613129	100	100	99.94	95.90
CTNNA2	114025	100	100	99.49	95.15
CTNND2	604275	94.56	92.55	90.44	84.97
DAB1	603448	100	100	100	100

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DAG1	128239	100	100	99.82	97.58
DCHS1	603057	100	100	100	99.83
DCX	300121	100	100	97.92	83.70
DDX3X	300160	100	100	100	97.21
DEPDC5	614191	100	100	99.65	97.73
DKC1	300126	100	99.10	97.39	86.54
DNMT3A	602769	100	100	94.09	74.73
DYNC1H1	600112	100	100	99.80	97.54
EIF2AK3	604032	100	97.98	94.32	90.79
EMG1	611531	100	100	100	100
EML1	602033	99.86	97.06	97.06	96.49
EOMES	604615	100	100	96.53	83.60
ERCC1	126380	100	99.34	95.56	68.39
ERCC2	126340	100	100	100	95.44
ERCC5	133530	100	100	98.52	91.56
ERCC6	609413	100	99.94	99.25	98.20
ERF	611888	100	100	97.56	91.33
ERMARD	615532	100	100	98.43	94.19
FAT4	612411	100	100	99.98	99.68
FIG4	609390	100	100	100	99.32
FKRP	606596	100	100	100	98.44
FKTN	607440	100	100	99.52	93.95
FLNA	300017	99.98	98.83	97.47	92.13
FLVCR2	610865	100	100	100	99.72
FOXC1	601090	97.11	90.56	84.87	73.23
FOXG1	164874	100	96.07	92.57	87.86
FRMD4A	616305	100	99.95	99.14	95.91
G3BP1	608431	100	98.92	95.77	84.73
G3BP2	No ID	100	100	100	98.26
GNAQ	600998	100	100	100	98.11
HNRNPK	600712	100	100	98.82	93.82
IBA57	615316	100	100	100	100
IER3IP1	609382	100	100	100	97.55
IFIH1	606951	100	100	100	99.12
INTS8	611351	99.91	98.40	95.50	87.06
ITSN1	602442	100	100	99.40	94.28
JAM3	606871	100	100	100	99.41
KATNB1	602703	100	100	100	99.55
KIF11	148760	100	99.50	96.77	89.01
KIF13B	607350	100	100	99.46	95.60
KIF2A	602591	100	100	98.28	88.42

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KIF5C	604593	100	98.83	95.34	87.27
KIF7	611254	99.84	96.33	91.96	81.11
KIFBP	609367	100	100	100	94.81
KNL1	609173	99.29	99.28	98.97	98.08
KPTN	615620	100	100	97.19	85.14
L1CAM	308840	100	100	99.08	86.61
LAMA1	150320	100	100	99.99	98.15
LAMA2	156225	100	100	99.89	97.74
LAMB1	150240	100	100	100	98.30
LAMC1	150290	100	99.74	98.81	95.38
LAMC3	604349	100	99.45	98.02	88.98
LARGE1	603590	100	100	99.45	96.92
LARP7	612026	100	99.19	94.94	79.55
MACF1	608271	100	100	99.71	97.26
MAP1A	600178	100	100	100	95.73
MCF2L	609499	100	100	98.56	93.29
MCPH1	607117	100	97.13	94.14	89.45
MDGA1	609626	100	99.91	98.56	92.54
MED13	603808	100	99.86	99.35	97.16
MED17	603810	100	100	99.45	92.64
MN1	156100	100	100	100	94.20
MPDZ	603785	98.36	98.36	98.36	97.32
MSI1	603328	94.22	89.75	81.73	62.41
MTOR	601231	100	99.87	99.52	97.16
MYCN	164840	100	100	97.68	91.49
NBN	602667	100	100	100	97.35
NCAPD2	615638	100	100	100	98.26
NCAPD3	609276	100	100	100	99.16
NCAPH	602332	100	99.09	94.73	92.37
NCAPH2	611230	100	100	99.46	96.08
NDE1	609449	100	100	96.78	82.83
NEDD4L	606384	100	99.80	97.43	93.46
NFIA	600727	100	95.90	95.14	88.28
NID1	131390	100	100	99.81	94.35
NIN	608684	100	100	99.22	96.48
NPRL2	607072	100	100	100	100
NPRL3	600928	100	100	99.80	87.95
NSDHL	300275	100	100	100	96.78
OCLN	602876	82.90	72.78	70.85	58.74
PAFAH1B1	601545	100	97.90	93.36	93.36
PAX6	607108	100	100	100	100

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PCDH12	605622	100	100	100	98.21
PCNT	605925	100	99.96	99.08	93.59
PHC1	602978	100	100	97.83	89.49
PHGDH	606879	100	100	100	96.57
PI4KA	600286	100	99.27	96.68	91.65
PIK3CA	171834	100	100	100	98.85
PIK3R2	603157	93.42	90.78	89.90	85.23
PLCH1	612835	100	100	100	98.32
PLK4	605031	100	100	100	97.35
PNKP	605610	100	100	98.86	93.95
POLR3B	614366	100	100	100	97.68
POMGNT1	606822	100	100	100	99.44
POMT1	607423	100	100	99.50	95.82
POMT2	607439	100	100	99.96	97.88
PPIL1	601301	100	100	100	100
PRUNE1	617413	100	100	100	100
PSAT1	610936	100	100	100	95.58
PSPH	172480	100	98.77	93.61	73.16
PTEN	601728	100	100	100	100
PTF1A	607194	100	97.85	85.01	50.63
PYCR2	616406	100	100	98.48	86.83
QARS1	603727	100	100	100	98.82
RAB18	602207	100	92.49	88.09	78.21
RAB3GAP1	602536	100	100	100	99.51
RAB3GAP2	609275	100	99.48	98.16	95.87
RAD50	604040	100	99.35	95.53	82.44
RARS2	611524	100	100	99.95	97.60
RBBP8	604124	100	100	99.41	95.13
RBM10	300080	99.99	97.66	94.82	80.06
RELN	600514	100	100	99.94	98.49
RHEB	601293	91.22	89.89	89.89	75.14
RNASEH2A	606034	100	100	100	95.93
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
RNASET2	612944	100	100	96.89	86.08
RNF125	610432	100	100	100	94.61
RNU4ATAC	601428	No coverage data			
ROBO3	608630	100	99.79	98.44	87.77
RTEL1	608833	100	100	99.83	95.32
RTTN	610436	100	100	99.96	96.53
RXYLT1	605862	100	100	100	100

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SAMHD1	606754	100	100	100	100
SCN3A	182391	100	100	100	99.27
SHANK3	606230	93.53	88.94	82.01	68.36
SHOC2	602775	100	100	100	98.74
SLC25A19	606521	100	100	99.91	97.04
SLC35A2	314375	100	100	86.85	83.56
SMPD4	610457	100	100	99.33	90.56
SNAP29	604202	100	100	100	99.43
SPEN	613484	100	100	100	99.73
SRPX2	300642	100	100	99.06	93.98
STAMBP	606247	100	100	100	99.70
STIL	181590	100	100	100	98.97
STRADA	608626	100	100	100	99.22
TAF2	604912	100	100	99.73	94.79
TBC1D20	611663	94.89	93.43	93.43	93.43
TBC1D24	613577	100	100	99.45	96.51
TBC1D7	612655	100	100	100	97.66
TBR1	604616	100	100	100	100
THG1L	618802	100	100	100	100
TMTC3	617218	100	100	98.93	89.87
TMX2	616715	100	100	100	100
TOGARAM1	617618	100	100	99.18	95.78
TRAIP	605958	100	100	100	100
TREX1	606609	100	100	100	100
TSC1	605284	100	100	100	99.95
TSC2	191092	100	100	100	97.97
TSEN54	608755	96.30	95.77	94.33	85.79
TUBA1A	602529	100	100	100	100
TUBA8	605742	98.41	98.41	98.41	98.41
TUBB	191130	100	100	100	98.37
TUBB2A	615101	100	99.61	94.56	75.58
TUBB2B	612850	100	100	99.61	81.13
TUBB3	602661	100	99.30	94.62	94.62
TUBB4A	602662	100	94.98	87.57	87.57
TUBG1	191135	100	100	100	100
TUBGCP4	609610	100	100	99.79	97.71
TUBGCP6	610053	100	100	100	98.75
VLDLR	192977	100	100	97.02	96.50
VPS13B	607817	99.37	98.22	97.00	90.85
VRK1	602168	100	100	100	96.88
WASHC5	610657	100	100	100	98.17

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WDR4	605924	100	100	97.65	84.66
WDR62	613583	100	100	99.32	94.62
WDR73	616144	100	100	100	100
WDR81	614218	100	99.68	98.13	91.95
YWHAE	605066	100	98.31	90.45	84.52
ZIC1	600470	100	100	98.72	93.11
ZIC2	603073	92.27	89.10	87.56	84.00
ZIC4	608948	100	100	100	100
ZNF462	617371	100	99.88	99.51	98.09
ZNF526	614387	100	100	100	100

- OMIM release used: 23-9-2021

- The statistics above are based on a set of 150 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