Whole Exome Sequencing
Gene package Neuronal migration 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 several (fragments of) genes involved in neuronal migration disorders (SALSA P061 Lissencephaly; MRC Holland). 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
ACTB	102630	192	100	100	100
ACTG1	102560	188	100	100	100
ADA2	607575	96	100	100	99
ADAR	146920	90	100	100	100
ADGRG1	604110	120	100	100	100
AGBL2	617345	51	97	94	85
AGTPBP1	606830	64	100	95	87
AKT1	164730	147	100	100	100
AKT3	611223	70	100	99	90
ANKLE2	616062	102	100	99	95
AP1S2	300629	40	100	87	62
AP3B2	602166	100	100	100	98
AP4B1	607245	81	100	100	100
AP4E1	607244	64	100	100	96
AP4M1	602296	130	100	100	99
AP4S1	607243	44	100	99	87
APC2	612034	128	100	97	95

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ARFGEF2	605371	83	100	100	96
ARNT2	606036	91	100	100	97
ARX	300382	46	89	79	68
ASNS	108370	73	100	100	92
ASPM	605481	71	100	100	97
ASXL1	612990	107	100	98	98
ATAD3A	612316	124	98	95	92
ATAD3B	612317	125	98	95	90
ATP6V0A2	611716	81	100	100	97
ATR	601215	91	100	99	92
ATRIP	606605	108	100	100	97
B3GALNT2	610194	59	100	100	97
B4GAT1	605517	154	100	100	100
BAP1	603089	121	100	100	100
CASK	300172	51	100	96	80
CCND2	123833	107	100	100	99
CDK5	123831	118	100	100	100
CDK5RAP2	608201	69	100	99	94
CDK6	603368	78	100	100	96
CENPJ	609279	74	100	100	98
CEP135	611423	83	100	99	91
CEP152	613529	67	100	98	93
CEP63	614724	72	100	99	92
CHMP1A	164010	102	100	100	100
CIT	605629	91	100	100	97
CLEC16A	611303	94	100	98	92
CLP1	608757	99	100	100	100
COL18A1	120328	143	100	100	97
COL4A1	120130	88	100	100	97
COL4A2	120090	102	100	100	99
COLGALT1	617531	110	97	90	86
CRADD	603454	139	100	100	100
CRB2	609720	115	100	100	100
CRPPA	614631	81	100	99	93
CSTB	601145	84	100	100	100
CTC1	613129	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CTNNA2	114025	82	100	100	98
CTNND2	604275	86	97	94	90
DAB1	603448	70	100	100	97
DAG1	128239	164	100	100	100
DCHS1	603057	128	100	100	100
DCX	300121	57	100	98	92
DDX3X	300160	76	100	100	98
DEPDC5	614191	83	100	100	97
DKC1	300126	53	100	98	89
DNMT3A	602769	117	100	100	99
DYNC1H1	600112	96	100	100	99
EIF2AK3	604032	72	100	99	94
EMG1	611531	78	100	100	100
EML1	602033	76	100	99	94
EOMES	604615	89	100	100	100
ERCC1	126380	71	100	100	94
ERCC2	126340	97	100	99	98
ERCC5	133530	85	100	100	98
ERCC6	609413	88	100	100	97
ERMARD	615532	67	100	100	96
FAT4	612411	90	100	100	99
FIG4	609390	59	100	99	94
FKRP	606596	143	100	100	100
FKTN	607440	85	100	100	100
FLNA	300017	110	100	100	100
FLVCR2	610865	140	100	100	99
FOXC1	601090	80	100	95	89
FRMD4A	616305	86	100	97	90
GNAQ	600998	94	100	100	100
HNRNPK	600712	46	96	84	66
IBA57	615316	124	100	100	97
IER3IP1	609382	83	100	100	77
IFIH1	606951	86	100	100	96
INTS8	611351	50	100	98	84
ITSN1	602442	66	100	97	91
JAM3	606871	70	100	100	97

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
KATNB1	602703	141	100	100	100
KIF11	148760	73	100	99	93
KIF2A	602591	84	100	99	90
KIF5C	604593	70	100	99	93
KIF7	611254	107	98	96	93
KIFBP	609367	76	100	100	98
KNL1	609173	60	100	99	95
KPTN	615620	150	100	100	100
L1CAM	308840	101	100	100	100
LAMA1	150320	87	100	100	97
LAMA2	156225	71	100	99	96
LAMB1	150240	94	100	100	97
LAMC1	150290	83	100	100	97
LAMC3	604349	130	100	100	99
LARGE1	603590	97	100	100	99
LARP7	612026	67	100	99	92
MACF1	608271	68	100	99	94
MAP1A	600178	121	100	100	100
MCF2L	609499	110	100	99	97
MCPH1	607117	85	94	94	92
MDGA1	609626	108	100	99	98
MED13	603808	57	100	98	92
MED17	603810	94	100	100	99
MN1	156100	151	100	100	100
MPDZ	603785	69	100	99	95
MTOR	601231	96	100	100	97
MYCN	164840	169	100	100	100
NBN	602667	68	100	99	88
NCAPD2	615638	90	100	100	98
NCAPD3	609276	65	100	98	92
NCAPH	602332	70	100	100	96
NCAPH2	611230	93	100	96	90
NDE1	609449	109	100	100	100
NEDD4L	606384	71	100	99	94
NFIA	600727	87	100	100	98
NID1	131390	114	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
NIN	608684	79	100	99	95
NPRL2	607072	119	100	100	100
NPRL3	600928	85	100	99	95
NSDHL	300275	68	100	100	93
OCLN	602876	61	96	84	79
PAFAH1B1	601545	82	100	96	90
PAX6	607108	73	100	100	96
PCDH12	605622	144	100	100	100
PCNT	605925	121	100	100	99
PHC1	602978	122	100	97	94
PI4KA	600286	114	100	99	96
PIK3CA	171834	85	100	99	96
PIK3R2	603157	89	95	93	90
PLK4	605031	62	100	99	91
PNKP	605610	104	100	100	98
POLR3B	614366	76	100	99	93
POMGNT1	606822	93	100	100	99
POMT1	607423	103	100	100	100
POMT2	607439	87	100	100	99
PRUNE1	617413	76	100	99	94
PTEN	601728	115	85	78	76
PTF1A	607194	136	100	100	94
PYCR2	616406	114	100	100	100
QARS1	603727	108	100	100	100
RAB18	602207	85	100	100	97
RAB3GAP1	602536	67	100	100	97
RAB3GAP2	609275	66	100	99	92
RAD50	604040	98	100	100	98
RARS2	611524	66	100	99	93
RBBP8	604124	57	100	99	92
RBM10	300080	81	100	96	88
RELN	600514	74	100	100	97
RHEB	601293	27	86	60	36
RNASEH2A	606034	109	100	100	100
RNASEH2B	610326	62	100	98	88
RNASEH2C	610330	323	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
RNASET2	612944	106	100	100	99
RNU4ATAC	601428	No coverage data			
ROBO3	608630	106	100	100	100
RTEL1	608833	131	100	100	99
RTTN	610436	71	100	99	94
RXYLT1	605862	90	100	100	94
SAMHD1	606754	64	100	99	88
SCN3A	182391	84	100	100	97
SHANK3	606230	117	99	93	86
SHOC2	602775	61	100	99	95
SLC25A19	606521	105	100	100	100
SLC35A2	314375	75	100	100	99
SMPD4	610457	112	100	100	98
SNAP29	604202	148	100	100	100
SRPX2	300642	59	100	99	95
STAMBP	606247	72	100	100	97
STIL	181590	65	100	100	97
STRADA	608626	96	100	100	98
TBC1D20	611663	74	100	93	93
TBC1D24	613577	158	100	100	100
TBC1D7	612655	55	100	98	85
TMTC3	617218	73	100	99	93
TMX2	616715	69	100	94	88
TRAIP	605958	98	100	100	100
TREX1	606609	234	100	100	100
TSC1	605284	167	100	100	100
TSC2	191092	184	100	100	100
TSEN54	608755	105	100	96	96
TUBA1A	602529	110	100	100	100
TUBA8	605742	118	100	100	100
TUBB	191130	198	100	99	97
TUBB2A	615101	99	99	82	74
TUBB2B	612850	119	100	87	78
TUBB3	602661	269	100	99	95
TUBB4A	602662	242	100	100	99
TUBG1	191135	190	100	100	100

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TUBGCP4	609610	71	100	98	94
TUBGCP6	610053	159	100	100	99
VLDLR	192977	70	100	100	97
VPS13B	607817	75	100	99	96
VRK1	602168	53	100	99	92
WASHC5	610657	62	100	100	94
WDR4	605924	117	100	100	100
WDR62	613583	139	100	100	100
WDR73	616144	152	100	100	97
WDR81	614218	155	100	100	100
YWHAE	605066	75	100	100	91
ZIC1	600470	255	100	100	100
ZIC2	603073	161	96	95	93
ZIC4	608948	132	100	100	99

- 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