Whole Exome Sequencing
Gene package Neuronal migration disorders, version 8, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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 Alissa Interpret 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	253	100	100	100
ACTG1	102560	233	100	100	100
ADA2	607575	157	100	100	100
ADAR	146920	163	100	100	100
ADGRG1	604110	233	100	100	100
AGBL2	617345	93	95	94	90
AGTPBP1	606830	63	95	86	73
AKT1	164730	284	100	100	100
AKT3	611223	53	100	90	75
ANKLE2	616062	152	99	97	92
AP1S2	300629	33	95	74	53
AP3B2	602166	182	100	100	100
AP4B1	607245	130	100	100	100
AP4E1	607244	66	97	87	75
AP4M1	602296	141	100	100	100
AP4S1	607243	50	100	96	78
APC2	612034	235	100	99	97
ARFGEF2	605371	114	100	97	93
ARNT2	606036	174	100	100	100
ARX	300382	100	92	89	84
ASNS	108370	82	100	98	91
ASPM	605481	64	94	87	75
ASXL1	612990	187	100	98	97
ATAD3A	612316	313	98	97	94
ATAD3B	612317	336	98	95	90
ATP6V0A2	611716	120	100	97	91

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ATR	601215	76	98	92	83
ATRIP	606605	189	100	98	92
B3GALNT2	610194	101	100	100	99
B4GAT1	605517	291	100	100	100
BAP1	603089	213	100	100	100
CASK	300172	60	96	86	70
CCND2	123833	152	100	100	100
CDK5	123831	223	100	100	100
CDK5RAP2	608201	113	99	94	90
CDK6	603368	137	98	94	93
CENPJ	609279	97	97	93	89
CEP135	611423	42	92	74	57
CEP152	613529	79	93	86	81
CEP63	614724	50	97	81	65
CHMP1A	164010	202	100	100	100
CIT	605629	131	100	100	99
CLEC16A	611303	141	100	99	96
CLP1	608757	213	100	100	100
COL18A1	120328	247	100	100	100
COL4A1	120130	121	100	99	96
COL4A2	120090	196	100	100	99
COLGALT1	617531	178	100	99	95
CRADD	603454	228	100	100	96
CRB2	609720	229	100	100	100
CRPPA	614631	76	97	84	77
CSTB	601145	132	100	100	100
CTC1	613129	174	100	100	100
CTNNA2	114025	106	100	98	93
CTNND2	604275	154	98	96	95
DAB1	603448	135	100	98	93
DAG1	128239	269	100	100	100
DCHS1	603057	292	100	100	100
DCX	300121	102	100	96	89
DDX3X	300160	56	100	98	91
DEPDC5	614191	157	100	100	98
DKC1	300126	73	100	93	88
DNMT3A	602769	198	100	100	100
DYNC1H1	600112	161	100	100	99
EIF2AK3	604032	95	97	93	90
EMG1	611531	153	100	100	100
EML1	602033	103	100	100	98
EOMES	604615	171	100	100	100
ERCC1	126380	143	100	100	100
ERCC2	126340	262	100	100	100
ERCC5	133530	87	99	96	89

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ERCC6	609413	128	100	100	98
ERMARD	615532	102	96	82	73
FAT4	612411	170	100	99	98
FIG4	609390	88	100	96	91
FKRP	606596	339	100	100	100
FKTN	607440	104	90	81	68
FLNA	300017	206	100	100	100
FLVCR2	610865	192	100	100	100
FOXC1	601090	137	100	96	93
FRMD4A	616305	174	100	100	100
GNAQ	600998	137	100	100	100
HNRNPK	600712	69	97	89	77
IBA57	615316	227	100	100	100
IER3IP1	609382	147	100	95	85
IFIH1	606951	70	99	91	80
INTS8	611351	53	92	77	67
ITSN1	602442	109	98	95	91
JAM3	606871	129	100	100	100
KATNB1	602703	334	100	100	100
KIF11	148760	46	95	80	64
KIF2A	602591	58	95	79	65
KIF5C	604593	99	99	93	87
KIF7	611254	224	100	100	98
KIFBP	609367	117	100	100	100
KNL1	609173	80	99	96	92
KPTN	615620	242	100	100	100
L1CAM	308840	164	100	100	100
LAMA1	150320	140	100	99	98
LAMA2	156225	96	100	98	93
LAMB1	150240	144	100	98	94
LAMC1	150290	152	100	98	95
LAMC3	604349	249	100	100	100
LARGE1	603590	204	100	100	100
LARP7	612026	37	95	79	62
MACF1	608271	104	100	99	97
MAP1A	600178	213	100	100	99
MCF2L	609499	230	100	100	100
MCPH1	607117	114	87	84	81
MDGA1	609626	223	100	100	100
MED13	603808	93	100	97	91
MED17	603810	82	97	87	68
MN1	156100	253	100	100	100
MPDZ	603785	104	100	99	95
MTOR	601231	147	100	100	100
MYCN	164840	275	100	100	100

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NBN	602667	57	98	83	71
NCAPD2	615638	136	100	100	100
NCAPD3	609276	113	100	100	97
NCAPH	602332	103	95	95	94
NCAPH2	611230	256	100	100	100
NDE1	609449	133	100	100	100
NEDD4L	606384	98	98	96	93
NFIA	600727	112	100	100	100
NID1	131390	181	100	100	100
NIN	608684	97	98	94	91
NPRL2	607072	317	100	100	100
NPRL3	600928	184	100	100	100
NSDHL	300275	88	100	100	95
OCLN	602876	100	85	81	69
PAFAH1B1	601545	72	86	86	85
PAX6	607108	267	100	100	100
PCDH12	605622	267	100	100	100
PCNT	605925	234	100	96	94
PHC1	602978	141	100	100	98
PI4KA	600286	153	100	100	99
PIK3CA	171834	69	100	95	89
PIK3R2	603157	272	96	94	93
PLK4	605031	77	98	90	83
PNKP	605610	246	100	100	100
POLR3B	614366	93	100	97	92
POMGNT1	606822	228	100	100	100
POMT1	607423	155	100	100	100
POMT2	607439	154	100	100	97
PRUNE1	617413	140	100	100	100
PTEN	601728	100	99	94	87
PTF1A	607194	138	100	97	93
PYCR2	616406	201	100	100	100
QARS1	603727	206	100	100	100
RAB18	602207	68	91	90	85
RAB3GAP1	602536	86	100	99	95
RAB3GAP2	609275	96	97	91	81
RAD50	604040	51	94	76	57
RARS2	611524	70	100	93	77
RBBP8	604124	53	99	89	79
RBM10	300080	130	100	100	100
RELN	600514	114	100	99	97
RHEB	601293	28	90	59	39
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	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	99	93	92	84
RNU4ATAC	601428	No coverage data			
ROBO3	608630	213	100	100	100
RTEL1	608833	260	100	100	100
RTTN	610436	74	98	94	87
RXYLT1	605862	75	99	91	82
SAMHD1	606754	97	100	99	93
SCN3A	182391	92	100	96	90
SHANK3	606230	246	99	98	97
SHOC2	602775	74	100	100	96
SLC25A19	606521	168	100	100	100
SLC35A2	314375	196	100	100	99
SMPD4	610457	243	100	99	97
SNAP29	604202	107	100	100	95
SRPX2	300642	102	100	100	100
STAMBP	606247	126	100	92	92
STIL	181590	192	100	99	95
STRADA	608626	154	100	100	96
TBC1D20	611663	149	100	100	96
TBC1D24	613577	390	100	100	100
TBC1D7	612655	94	95	85	85
TBR1	604616	270	100	100	100
TMTC3	617218	47	96	79	62
TMX2	616715	147	100	100	100
TRAIP	605958	175	100	100	100
TREX1	606609	313	100	100	100
TSC1	605284	153	100	100	100
TSC2	191092	314	100	100	100
TSEN54	608755	201	100	100	98
TUBA1A	602529	219	100	100	100
TUBA8	605742	185	100	100	100
TUBB	191130	186	100	100	100
TUBB2A	615101	109	100	90	81
TUBB2B	612850	113	100	90	80
TUBB3	602661	330	100	100	97
TUBB4A	602662	233	100	100	100
TUBG1	191135	334	100	100	100
TUBGCP4	609610	122	100	100	98
TUBGCP6	610053	356	100	100	100
VLDLR	192977	120	100	100	98
VPS13B	607817	98	99	95	89
VRK1	602168	56	100	89	72
WASHC5	610657	82	100	94	87
WDR4	605924	188	100	100	100
WDR62	613583	214	100	100	100

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WDR73	616144	194	100	100	100
WDR81	614218	293	100	100	100
YWHAE	605066	85	87	85	82
ZIC1	600470	322	100	100	100
ZIC2	603073	171	95	93	91
ZIC4	608948	291	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