

Whole Exome Sequencing

Gene package Epilepsy, version 5.1, 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. 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).



Dept. Clinical Genetics

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
AARS1	601065	161	100	100	100
ABAT	137150	142	100	100	100
ABCC8	600509	242	100	100	100
ACTB	102630	253	100	100	100
ACTL6B	612458	238	100	100	100
ACY1	104620	215	100	100	100
ADSL	608222	178	100	100	100
ALDH7A1	107323	105	100	100	95
ALG1	605907	126	100	100	99
ALG11	613666	109	100	100	95
ALG13	300776	67	98	92	81
ALG3	608750	213	100	100	100
ALG6	604566	49	90	81	67
AMACR	604489	129	100	100	99
AMT	238310	225	100	100	100
ANKRD11	611192	247	100	99	97
AP3B2	602166	182	100	100	100
ARHGEF9	300429	93	100	100	99
ARID1B	614556	166	100	100	98
ARV1	611647	110	100	97	88
ARX	300382	100	92	89	84
ASAH1	613468	73	98	92	81
ASL	608310	294	100	100	100
ATAD1	614452	65	94	93	87
ATP1A2	182340	189	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ATP1A3	182350	221	100	100	100
ATP6AP2	300556	54	100	93	81
ATP7A	300011	59	99	94	81
ATRX	300032	37	92	74	52
AUTS2	607270	204	100	99	98
BOLA3	613183	63	100	98	94
BRAT1	614506	243	100	100	100
BTD	609019	195	100	100	100
CACNA1A	601011	139	100	100	99
CACNA1E	601013	174	100	100	100
CACNA2D2	607082	193	98	96	94
CACNB4	601949	176	100	100	99
CASK	300172	60	96	86	70
CDKL5	300203	74	97	92	86
CERS1	606919	150	83	77	77
CERT1	604677	95	96	92	89
CHD2	602119	82	98	92	84
CHRNA2	118502	314	100	100	100
CHRNA4	118504	386	100	100	99
CHRN2	118507	245	100	100	100
CLCN4	302910	141	100	100	98
CLDN16	603959	120	100	100	100
CLDN19	610036	369	100	100	100
CLN3	607042	211	100	100	100
CLN5	608102	113	100	93	89
CLN6	606725	264	100	100	100
CLN8	607837	195	100	100	100
CNKSR2	300724	49	96	82	65
CNNM2	607803	209	100	100	97
CNTN2	190197	247	100	100	100
CNTNAP2	604569	119	100	100	99
COA8	616003	91	100	94	80
COL4A1	120130	121	100	99	96
COQ2	609825	79	98	92	82
COQ4	612898	176	100	100	97
COQ8A	606980	250	100	100	100
CPA6	609562	99	96	94	91
CPS1	608307	101	100	100	99
CPT2	600650	187	100	100	100
CSNK2B	115441	123	100	100	100
CSTB	601145	132	100	100	100
CTNND2	604275	154	98	96	95
CTSD	116840	278	100	100	100
CTSF	603539	232	100	98	96

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CUL4B	300304	43	90	77	62
D2HGDH	609186	269	100	100	100
DCX	300121	102	100	96	89
DENND5A	617278	136	100	100	98
DEPDC5	614191	157	100	100	98
DLAT	608770	131	100	99	92
DNAJC5	611203	258	100	100	100
DNM1	602377	172	100	100	99
DOCK7	615730	72	99	94	87
DPAGT1	191350	151	100	100	100
DPM1	603503	79	98	89	73
DPM2	603564	164	100	100	100
DPYD	612779	73	94	92	86
DYNC1H1	600112	161	100	100	99
DYRK1A	600855	111	100	97	92
EEF1A2	602959	218	100	100	100
EGF	131530	110	99	95	91
EHMT1	607001	176	99	99	98
EPM2A	607566	164	90	86	84
FA2H	611026	182	100	100	100
FARS2	611592	221	100	100	100
FASN	600212	322	100	100	100
FGD1	300546	126	100	100	100
FLNA	300017	206	100	100	100
FOLR1	136430	146	100	100	100
FOXG1	164874	424	100	97	94
FOXRED1	613622	260	100	100	100
FRRS1L	604574	75	100	92	80
FXYD2	601814	241	100	100	100
GABRA1	137160	96	100	99	97
GABRA2	137140	69	99	93	83
GABRA3	305660	73	100	98	92
GABRB2	600232	120	100	100	99
GABRB3	137192	160	100	100	100
GABRE	300093	86	100	97	91
GABRG2	137164	98	100	99	93
GAMT	601240	284	100	100	100
GCK	138079	256	100	100	100
GCSH	238330	43	100	85	69
GLDC	238300	118	100	100	98
GLRA1	138491	154	100	100	100
GLRB	138492	74	95	91	79
GLUD1	138130	123	100	98	93
GNAO1	139311	173	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
GOSR2	604027	139	100	100	100
GPC3	300037	86	100	98	92
GPHN	603930	87	100	96	94
GRIA3	305915	62	98	91	79
GRIK2	138244	66	99	93	86
GRIN1	138249	259	100	100	100
GRIN2A	138253	408	100	100	99
GRIN2B	138252	237	100	100	99
GRIN2D	602717	143	97	88	83
GRN	138945	336	100	100	100
HADH	601609	145	100	100	98
HCFC1	300019	207	100	100	100
HCN1	602780	193	100	99	97
HDAC4	605314	227	100	99	97
HLCS	609018	163	100	100	99
HNRNPU	602869	88	95	94	92
HSD17B10	300256	123	100	100	100
HSD17B4	601860	78	93	91	84
HUWE1	300697	90	100	98	93
IDH2	147650	236	100	100	100
IER3IP1	609382	147	100	95	85
IFIH1	606951	70	99	91	80
INTS8	611351	53	92	77	67
IQSEC2	300522	128	100	99	98
IRF2BPL	611720	325	100	100	98
JAM3	606871	129	100	100	100
KANSL1	612452	188	100	100	100
KCNA1	176260	224	100	100	100
KCNA2	176262	209	100	100	100
KCNB1	600397	279	100	100	100
KCNC1	176258	275	100	100	100
KCND3	605411	350	100	100	99
KCNH1	603305	163	100	100	99
KCNJ10	602208	271	100	100	100
KCNJ11	600937	382	100	100	100
KCNMA1	600150	122	100	98	96
KCNQ2	602235	274	100	100	100
KCNQ3	602232	186	100	100	98
KCNQ5	607357	107	100	99	93
KCNT1	608167	236	100	100	100
KCTD7	611725	259	100	100	100
KDM5C	314690	130	100	100	99
KMT2A	159555	118	100	100	98
KPNA7	614107	159	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
KPTN	615620	242	100	100	100
LGI1	604619	84	98	93	84
LIAS	607031	78	95	93	87
MBD5	611472	141	100	99	97
MDH2	154100	167	100	100	100
MECP2	300005	200	100	100	99
MED12	300188	99	100	100	98
MEF2C	600662	157	100	100	99
MFSD8	611124	82	99	93	83
MLC1	605908	181	100	100	95
MOCS1	603707	198	100	100	100
MOCS2	603708	81	100	100	97
MPDU1	604041	164	100	100	100
MTHFR	607093	191	100	100	100
MTOR	601231	147	100	100	100
NAPB	611270	101	99	90	84
NBEA	604889	68	98	93	82
NDUFA1	300078	106	100	100	100
NDUFA11	612638	243	100	100	100
NDUFAF1	606934	87	100	100	98
NDUFAF2	609653	77	100	99	90
NDUFAF3	612911	230	100	100	100
NDUFAF4	611776	61	90	53	38
NDUFAF5	612360	73	96	94	87
NDUFB3	603839	72	100	100	100
NDUFB9	601445	153	100	100	98
NDUFS1	157655	90	100	99	96
NDUFS2	602985	143	100	100	100
NDUFS3	603846	180	100	100	100
NDUFS4	602694	88	100	100	98
NDUFS6	603848	99	100	100	95
NDUFV1	161015	257	100	100	100
NDUFV2	600532	68	98	90	77
NECAP1	611623	150	100	100	100
NEDD4L	606384	98	98	96	93
NEXMIF	300524	94	100	100	100
NGLY1	610661	70	100	97	89
NHLRC1	608072	312	100	100	100
NPRL2	607072	317	100	100	100
NPRL3	600928	184	100	100	100
NRXN1	600565	190	100	99	98
NSDHL	300275	88	100	100	95
NUBPL	613621	108	100	95	88
OFD1	300170	51	97	80	64

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OPHN1	300127	71	100	99	91
PAK3	300142	41	94	88	73
PC	608786	344	100	100	100
PCDH19	300460	221	100	100	98
PDHA1	300502	78	100	94	87
PDHB	179060	104	100	100	99
PDP1	605993	194	100	100	100
PDX1	600733	153	100	100	100
PET100	614770	178	100	100	100
PEX1	602136	80	95	94	90
PEX10	602859	266	100	100	100
PEX12	601758	131	100	100	100
PEX13	601789	113	100	100	100
PEX14	601791	188	100	100	100
PEX16	603360	212	100	100	100
PEX19	600279	205	100	100	100
PEX26	608666	151	100	100	100
PEX3	603164	60	100	97	88
PEX5	600414	172	100	100	100
PEX6	601498	253	100	100	99
PGAP1	611655	42	92	74	56
PGAP3	611801	208	100	100	100
PHF6	300414	22	86	52	22
PHGDH	606879	199	100	100	100
PIGA	311770	62	100	99	86
PIGN	606097	50	92	79	65
PIGO	614730	253	100	100	100
PIGT	610272	194	100	100	100
PLA2G6	603604	287	100	100	100
PLCB1	607120	80	98	94	87
PLP1	300401	86	100	98	92
PMM2	601785	115	100	100	99
PNKP	605610	246	100	100	100
PNPO	603287	181	100	100	100
POLG	174763	188	100	100	100
PPP2R1A	605983	241	100	100	100
PPP3CA	114105	97	100	99	93
PPT1	600722	145	100	100	100
PQBP1	300463	124	100	100	100
PRICKLE1	608500	107	100	100	99
PRICKLE2	608501	239	100	100	100
PRIMA1	613851	72	89	85	83
PRRT2	614386	179	100	100	98
PSAT1	610936	103	100	98	90

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PSPH	172480	97	96	83	78
PURA	600473	481	100	100	100
PYCR2	616406	201	100	100	100
QARS1	603727	206	100	100	100
RAB39B	300774	156	100	100	99
RAI1	607642	405	100	100	100
RANBP2	601181	91	95	90	85
RARS2	611524	70	100	93	77
RELN	600514	114	100	99	97
RNASEH2A	606034	265	100	100	100
RNASEH2B	610326	60	100	99	89
RNASEH2C	610330	261	100	100	100
ROGDI	614574	222	100	99	97
RPS6KA3	300075	37	96	80	55
RRM2B	604712	117	100	100	98
SAMHD1	606754	97	100	99	93
SCARB2	602257	79	100	97	91
SCN1A	182389	97	100	97	91
SCN1B	600235	224	100	100	100
SCN2A	182390	93	98	94	86
SCN8A	600702	137	100	100	98
SHANK3	606230	246	99	98	97
SIK1	605705	309	100	100	100
SLC12A5	606726	174	100	100	100
SLC13A5	608305	194	100	100	100
SLC16A1	600682	117	100	100	99
SLC19A3	606152	108	100	100	100
SLC1A3	600111	106	100	99	95
SLC25A1	190315	204	100	100	100
SLC25A15	603861	152	100	100	100
SLC25A22	609302	317	100	100	100
SLC2A1	138140	354	100	100	100
SLC35A2	314375	196	100	100	99
SLC6A1	137165	177	100	100	98
SLC6A5	604159	154	100	100	100
SLC6A8	300036	134	100	100	97
SLC9A6	300231	89	98	88	81
SMARCA2	600014	95	100	98	96
SMC1A	300040	96	100	96	95
SMS	300105	61	100	93	86
SNAP25	600322	96	100	100	96
SON	182465	170	100	98	94
SPTAN1	182810	147	100	99	98
SRPX2	300642	102	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
ST3GAL3	606494	156	100	100	100
ST3GAL5	604402	100	100	93	92
STX1B	601485	139	100	100	100
STXBP1	602926	126	100	100	100
SUOX	606887	254	100	100	100
SYN1	313440	151	100	100	100
SYNGAP1	603384	201	100	98	98
SYNJ1	604297	83	98	95	90
SYP	313475	110	100	100	99
SZT2	615463	206	100	100	100
TANGO2	616830	253	100	100	100
TBC1D24	613577	390	100	100	100
TBCE	604934	69	100	96	87
TBCK	616899	56	95	88	76
TCF4	602272	145	100	99	97
TDP2	605764	121	89	88	84
TPP1	607998	223	100	100	100
TREX1	606609	313	100	100	100
TRIO	601893	159	99	99	97
TRPM6	607009	192	98	97	96
TSC1	605284	153	100	100	100
TSC2	191092	314	100	100	100
TSEN54	608755	201	100	100	98
TUBA1A	602529	219	100	100	100
TUBB2A	615101	109	100	90	81
TUBG1	191135	334	100	100	100
UBA5	610552	52	93	78	66
UBE2A	312180	96	100	98	80
UBE3A	601623	79	98	89	83
UGDH	603370	88	99	91	84
UGP2	191760	64	99	91	80
WDR45	300526	161	100	100	100
WWOX	605131	128	100	94	94
XK	314850	107	100	100	100
YWHAG	605356	198	100	100	100
ZDHC9	300646	77	100	100	98
ZEB2	605802	289	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 (± 10 bp flanking introns) of the longest transcript

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

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
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