Whole Exome Sequencing
Gene package Epilepsy, version 5, 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. 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
AARS1	601065	89	100	100	96
ABAT	137150	100	100	100	96
ABCC8	600509	105	100	100	100
ACTB	102630	192	100	100	100
ACTL6B	612458	124	100	100	100
ACY1	104620	109	100	100	100
ADSL	608222	89	100	100	96
ALDH7A1	107323	69	100	100	91
ALG1	605907	61	91	79	73
ALG11	613666	73	100	100	99
ALG13	300776	50	100	97	83
ALG3	608750	95	100	100	100
ALG6	604566	73	100	100	96
AMACR	604489	96	100	100	98
AMT	238310	130	100	100	100
ANKRD11	611192	112	100	98	96
AP3B2	602166	100	100	100	98

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ARHGEF9	300429	50	100	99	85
ARID1B	614556	102	100	100	98
ARV1	611647	92	99	89	86
ARX	300382	46	89	79	68
ASAH1	613468	67	100	100	93
ASL	608310	117	100	100	99
ATAD1	614452	42	100	95	76
ATP1A2	182340	130	100	100	100
ATP1A3	182350	144	100	100	100
ATP6AP2	300556	46	100	91	64
ATP7A	300011	49	100	98	84
ATRX	300032	40	100	94	78
AUTS2	607270	122	100	100	98
BOLA3	613183	69	100	100	91
BRAT1	614506	125	100	100	100
BTD	609019	89	100	100	100
CACNA1A	601011	87	100	98	93
CACNA1E	601013	106	100	99	97
CACNA2D2	607082	102	99	98	95
CACNB4	601949	57	100	100	94
CASK	300172	51	100	96	80
CDKL5	300203	58	100	97	87
CERS1	606919	94	90	80	77
CERT1	604677	62	100	96	85
CHD2	602119	72	100	100	96
CHRNA2	118502	170	100	100	100
CHRNA4	118504	138	100	100	96
CHRNB2	118507	144	100	100	100
CLCN4	302910	80	100	100	98
CLDN16	603959	73	100	100	97
CLDN19	610036	149	100	100	100
CLN3	607042	111	100	100	100
CLN5	608102	70	100	100	100
CLN6	606725	120	100	100	99
CLN8	607837	129	100	100	100
CNKSR2	300724	56	100	97	87

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CNNM2	607803	161	100	100	99
CNTN2	190197	113	100	100	100
CNTNAP2	604569	73	100	100	99
COA8	616003	69	100	100	92
COL4A1	120130	88	100	100	97
COQ2	609825	66	100	100	94
COQ4	612898	110	100	100	100
COQ8A	606980	129	100	100	100
CPA6	609562	79	100	99	94
CPS1	608307	61	100	100	96
CPT2	600650	106	100	100	99
CSNK2B	115441	90	100	100	99
CSTB	601145	84	100	100	100
CTNND2	604275	86	97	94	90
CTSD	116840	137	100	100	100
CTSF	603539	94	100	94	91
CUL4B	300304	52	100	96	82
D2HGDH	609186	140	100	100	100
DCX	300121	57	100	98	92
DENND5A	617278	64	100	99	94
DEPDC5	614191	83	100	100	97
DLAT	608770	74	100	98	90
DNAJC5	611203	217	100	100	100
DNM1	602377	138	100	100	95
DOCK7	615730	56	100	99	93
DPAGT1	191350	76	100	100	100
DPM1	603503	79	94	89	85
DPM2	603564	76	100	100	100
DPYD	612779	66	100	99	95
DYNC1H1	600112	96	100	100	99
DYRK1A	600855	75	100	100	98
EEF1A2	602959	169	100	100	100
EGF	131530	74	100	100	97
EHMT1	607001	126	99	99	99
EPM2A	607566	86	89	87	85
FA2H	611026	83	100	99	91

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
FARS2	611592	111	100	100	100
FASN	600212	130	100	100	97
FGD1	300546	71	100	98	95
FLNA	300017	110	100	100	100
FOLR1	136430	111	100	100	100
FOXG1	164874	127	97	90	84
FOXRED1	613622	91	100	100	100
FRRS1L	604574	50	85	79	70
FXYD2	601814	101	100	100	100
GABRA1	137160	76	100	100	100
GABRA2	137140	86	100	100	96
GABRA3	305660	44	97	88	69
GABRB2	600232	70	100	100	99
GABRB3	137192	95	100	100	98
GABRE	300093	56	100	94	81
GABRG2	137164	73	94	92	91
GAMT	601240	85	100	99	95
GCK	138079	127	100	100	100
GCSH	238330	93	100	89	62
GLDC	238300	67	100	98	91
GLRA1	138491	85	100	100	96
GLRB	138492	63	100	100	98
GLUD1	138130	99	100	97	91
GNAO1	139311	98	100	100	99
GOSR2	604027	66	100	95	90
GPC3	300037	53	100	99	91
GPHN	603930	68	100	100	98
GRIA3	305915	51	100	98	88
GRIK2	138244	94	100	100	98
GRIN1	138249	132	100	100	99
GRIN2A	138253	103	100	100	100
GRIN2B	138252	122	100	100	99
GRIN2D	602717	68	88	74	69
GRN	138945	155	100	100	100
HADH	601609	77	100	100	100
HCFC1	300019	79	100	97	92

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HCN1	602780	79	100	100	98
HDAC4	605314	112	100	100	100
HLCS	609018	98	100	100	99
HNRNPU	602869	81	100	100	95
HSD17B10	300256	76	100	100	100
HSD17B4	601860	62	100	98	93
HUWE1	300697	53	100	96	83
IDH2	147650	105	100	100	100
IER3IP1	609382	83	100	100	77
IFIH1	606951	86	100	100	96
INTS8	611351	50	100	98	84
IQSEC2	300522	66	98	92	85
IRF2BPL	611720	137	100	98	95
JAM3	606871	70	100	100	97
KANSL1	612452	85	100	100	97
KCNA1	176260	126	100	100	100
KCNA2	176262	108	100	100	100
KCNB1	600397	123	100	100	100
KCNC1	176258	138	100	100	99
KCND3	605411	167	100	100	99
KCNH1	603305	114	100	100	98
KCNJ10	602208	171	100	100	100
KCNJ11	600937	128	100	100	100
KCNMA1	600150	88	100	100	96
KCNQ2	602235	131	100	100	100
KCNQ3	602232	108	100	100	96
KCNQ5	607357	85	100	98	95
KCNT1	608167	120	100	99	99
KCTD7	611725	159	100	100	100
KDM5C	314690	88	100	99	96
KMT2A	159555	71	100	100	98
KPNA7	614107	89	100	100	100
KPTN	615620	150	100	100	100
LGI1	604619	83	100	100	98
LIAS	607031	78	100	100	95
MBD5	611472	81	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
MDH2	154100	103	100	100	100
MECP2	300005	113	100	100	96
MED12	300188	68	100	100	97
MEF2C	600662	93	100	99	96
MFSD8	611124	68	100	100	95
MLC1	605908	79	100	100	94
MOCS1	603707	105	100	100	99
MOCS2	603708	67	100	100	96
MPDU1	604041	87	100	100	99
MTHFR	607093	99	100	100	99
MTOR	601231	96	100	100	97
NAPB	611270	59	100	100	94
NBEA	604889	70	99	97	91
NDUFA1	300078	110	100	100	100
NDUFA11	612638	117	100	100	100
NDUFAF1	606934	63	100	100	95
NDUFAF2	609653	83	100	92	78
NDUFAF3	612911	130	100	100	100
NDUFAF4	611776	95	100	100	100
NDUFAF5	612360	70	100	100	96
NDUFB3	603839	31	100	87	55
NDUFB9	601445	102	100	100	97
NDUFS1	157655	69	100	100	94
NDUFS2	602985	79	100	100	99
NDUFS3	603846	112	100	100	100
NDUFS4	602694	83	100	100	99
NDUFS6	603848	70	100	100	100
NDUFV1	161015	129	100	100	100
NDUFV2	600532	48	100	95	80
NECAP1	611623	82	100	100	100
NEDD4L	606384	71	100	99	94
NEXMIF	300524	48	100	100	95
NGLY1	610661	78	100	100	97
NHLRC1	608072	144	100	100	100
NPRL2	607072	119	100	100	100
NPRL3	600928	85	100	99	95

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NRXN1	600565	104	100	100	97
NSDHL	300275	68	100	100	93
NUBPL	613621	60	100	100	97
OFD1	300170	43	100	95	75
OPHN1	300127	52	100	96	84
PAK3	300142	48	100	96	83
PC	608786	141	100	100	100
PCDH19	300460	113	100	100	97
PDHA1	300502	49	100	94	84
PDHB	179060	65	100	100	95
PDP1	605993	87	100	100	100
PDX1	600733	79	100	100	89
PET100	614770	85	92	66	66
PEX1	602136	62	100	99	94
PEX10	602859	100	100	100	97
PEX12	601758	67	100	100	94
PEX13	601789	69	100	100	99
PEX14	601791	142	100	100	100
PEX16	603360	134	100	96	94
PEX19	600279	67	100	100	98
PEX26	608666	118	100	100	100
PEX3	603164	57	100	100	95
PEX5	600414	111	100	100	100
PEX6	601498	107	100	99	95
PGAP1	611655	65	100	99	90
PGAP3	611801	115	100	100	100
PHF6	300414	51	100	96	82
PHGDH	606879	138	100	100	100
PIGA	311770	67	100	100	97
PIGN	606097	63	100	99	89
PIGO	614730	126	100	100	100
PIGT	610272	138	100	100	100
PLA2G6	603604	118	100	100	99
PLCB1	607120	60	100	99	94
PLP1	300401	93	100	99	97
PMM2	601785	72	100	100	97

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PNKP	605610	104	100	100	98
PNPO	603287	69	100	100	94
POLG	174763	114	100	100	99
PPP2R1A	605983	130	100	100	100
PPP3CA	114105	76	100	99	90
PPT1	600722	84	100	100	100
PQBP1	300463	100	100	100	100
PRICKLE1	608500	76	100	100	98
PRICKLE2	608501	113	100	100	100
PRIMA1	613851	60	88	85	82
PRRT2	614386	123	100	100	100
PSAT1	610936	64	100	100	97
PSPH	172480	51	100	96	84
PURA	600473	206	100	100	100
PYCR2	616406	114	100	100	100
QARS1	603727	108	100	100	100
RAB39B	300774	59	100	100	100
RAI1	607642	187	100	100	100
RANBP2	601181	112	100	100	99
RARS2	611524	66	100	99	93
RELN	600514	74	100	100	97
RNASEH2A	606034	109	100	100	100
RNASEH2B	610326	62	100	98	88
RNASEH2C	610330	323	100	100	100
ROGDI	614574	90	100	98	93
RPS6KA3	300075	46	100	92	70
RRM2B	604712	90	100	100	100
SAMHD1	606754	64	100	99	88
SCARB2	602257	89	100	100	98
SCN1A	182389	92	100	100	99
SCN1B	600235	169	100	97	93
SCN2A	182390	95	100	100	97
SCN8A	600702	110	100	100	98
SHANK3	606230	117	99	93	86
SIK1	605705	125	100	100	99
SLC12A5	606726	107	100	100	99

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
SLC13A5	608305	126	100	100	98
SLC16A1	600682	83	100	100	99
SLC19A3	606152	73	100	100	98
SLC1A3	600111	99	100	100	100
SLC25A1	190315	102	100	100	100
SLC25A15	603861	132	100	100	98
SLC25A22	609302	122	100	100	100
SLC2A1	138140	127	100	100	100
SLC35A2	314375	75	100	100	99
SLC6A1	137165	115	100	100	100
SLC6A5	604159	85	100	100	96
SLC6A8	300036	103	100	98	95
SLC9A6	300231	64	100	96	84
SMARCA2	600014	94	98	98	96
SMC1A	300040	71	100	100	97
SMS	300105	42	95	89	68
SNAP25	600322	84	100	100	98
SON	182465	85	99	96	90
SPTAN1	182810	95	100	100	98
SRPX2	300642	59	100	99	95
ST3GAL3	606494	79	100	100	96
ST3GAL5	604402	55	99	92	84
STX1B	601485	131	100	100	99
STXBP1	602926	76	100	100	99
SUOX	606887	136	100	100	100
SYN1	313440	60	100	99	85
SYNGAP1	603384	158	98	98	98
SYNJ1	604297	62	100	99	92
SYP	313475	69	100	100	98
SZT2	615463	117	100	100	100
TANGO2	616830	96	100	100	99
TBC1D24	613577	158	100	100	100
TBCE	604934	59	100	97	88
TBCK	616899	53	100	99	91
TCF4	602272	73	100	100	96
TDP2	605764	120	100	100	99

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TPP1	607998	97	100	100	100
TREX1	606609	234	100	100	100
TRIO	601893	100	99	99	97
TRPM6	607009	75	100	100	96
TSC1	605284	167	100	100	100
TSC2	191092	184	100	100	100
TSEN54	608755	105	100	96	96
TUBA1A	602529	110	100	100	100
TUBB2A	615101	99	99	82	74
TUBG1	191135	190	100	100	100
UBA5	610552	46	100	91	67
UBE2A	312180	57	100	99	81
UBE3A	601623	61	100	100	96
UGDH	603370	46	100	98	87
UGP2	191760	49	100	99	91
WDR45	300526	92	100	100	100
WWOX	605131	92	100	100	100
XK	314850	62	100	100	96
YWHAG	605356	121	100	100	100
ZDHHC9	300646	44	100	97	83
ZEB2	605802	85	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