

Whole Exome Sequencing

Gene package Epilepsy, version 8, 25-2-2022



Technical information

DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number 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 annotated and filtered with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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 $\geq 50x$
AARS1	601065	100	100	100	99.80
ABAT	137150	93.95	93.95	92.80	82.33
ABCC8	600509	100	100	100	98.90
ACTB	102630	100	100	100	99.18
ACTL6B	612458	100	100	99.42	90.28
ACY1	104620	100	100	99.67	89.10
ADSL	608222	98.31	98.31	98.31	98.31
ALDH7A1	107323	100	100	100	99.34
ALG1	605907	100	98.64	95.61	86.08
ALG11	613666	100	100	100	99.65
ALG13	300776	100	100	100	97.07
ALG3	608750	100	100	98.86	94.65
ALG6	604566	100	97.83	95.78	95.06
AMACR	604489	100	100	100	100
AMT	238310	100	100	100	97.08
ANKRD11	611192	100	99.03	97.64	94.15
AP2M1	601024	100	100	100	99.57
AP3B2	602166	100	100	90.63	74.57

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ARHGEF9	300429	96.19	96.19	96.00	91.27
ARID1B	614556	97.28	93.67	88.64	78.80
ARV1	611647	100	100	100	100
ARX	300382	84.88	79.12	68.56	50.95
ASAH1	613468	100	100	99.41	95.80
ASL	608310	100	100	100	96.47
ATAD1	614452	100	100	100	99.60
ATP1A2	182340	100	100	99.93	95.75
ATP1A3	182350	100	100	99.45	94.42
ATP6AP2	300556	100	100	99.59	93.78
ATP7A	300011	100	100	99.70	97.72
ATRX	300032	100	100	99.00	93.02
AUTS2	607270	100	99.61	95.60	80.85
BOLA3	613183	100	100	100	81.55
BRAT1	614506	100	96.68	92.21	88.41
BTD	609019	100	100	100	100
CACNA1A	601011	100	99.17	95.07	79.63
CACNA1B	601012	97.00	95.50	93.70	87.69
CACNA1E	601013	100	99.94	99.68	98.66
CACNA1G	604065	100	99.43	98.45	93.42
CACNA2D2	607082	94.66	94.47	92.75	83.19
CACNB4	601949	100	96.77	95	92.53
CAD	114010	100	100	99.90	96.49
CASK	300172	100	99.97	97.02	90.61
CDKL5	300203	100	99.94	98.62	93.38
CERS1	606919	100	100	100	99.02
CERT1	604677	100	100	100	98.91
CHD2	602119	100	99.69	98.24	91.83
CHD5	610771	99.96	97.99	94.30	83.61
CHRNA2	118502	100	100	100	96.19
CHRNA4	118504	100	96.33	95.20	94.85
CHRN2	118507	100	100	98.68	94.13
CLCN4	302910	100	100	99.10	93.72
CLCN6	602726	100	100	100	97.72
CLDN16	603959	100	100	100	100
CLDN19	610036	100	100	100	100
CLN3	607042	100	98.64	91.76	88.17
CLN5	608102	93.14	93.14	88.00	73.62
CLN6	606725	100	100	100	96.55
CLN8	607837	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
CNKS2R2	300724	100	99.92	98.83	88.92
CNNM2	607803	100	100	100	98.11
CNTN2	190197	100	100	100	97.19
CNTNAP1	602346	100	100	99.95	94.05
CNTNAP2	604569	100	100	99.59	97.84
COA8	616003	100	100	100	100
COL4A1	120130	99.39	97.25	93.74	85.37
COQ2	609825	100	100	99.68	89.02
COQ4	612898	100	93.69	80.43	63.74
COQ8A	606980	100	100	100	97.43
CPS1	608307	99.28	99.28	99.28	99.15
CPT2	600650	100	100	99.23	96.96
CSNK2B	115441	100	100	100	100
CSTB	601145	100	100	99.58	75.71
CTNND2	604275	94.24	92.38	90.15	84.22
CTSD	116840	100	93.79	93.79	90.47
CTSF	603539	95.77	88.58	86.39	83.35
CUL4B	300304	100	100	99.37	94.11
CUX1	116896	98.87	96.78	93.44	86.15
D2HGDH	609186	100	100	98.48	95.27
DCX	300121	100	100	98.63	88.82
DEAF1	602635	96.67	89.74	83.62	77.57
DENND5A	617278	100	100	99.43	96.20
DEPDC5	614191	100	99.98	99.37	97.07
DHDDS	608172	100	100	100	93.45
DLAT	608770	100	99.98	98.91	96.09
DNAJC5	611203	100	100	98.81	82.05
DNM1	602377	99.90	96.55	93.03	88.31
DOCK7	615730	100	99.62	98.23	96.12
DPAGT1	191350	100	100	100	97.76
DPM1	603503	100	100	100	99.38
DPM2	603564	100	100	100	89.16
DPYD	612779	100	100	100	100
DYNC1H1	600112	100	100	99.77	97.46
DYRK1A	600855	100	100	100	98.70
EEF1A2	602959	100	99.74	98.79	88.55
EGF	131530	100	100	99.73	96.57
EHMT1	607001	99.08	99.04	97.01	92.86
EPM2A	607566	85.89	81.07	79.79	76.89
FA2H	611026	100	96.58	89.53	72.53

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FARS2	611592	100	100	100	100
FASN	600212	100	99.95	98.91	94.66
FBXO28	609100	100	100	95.95	81.28
FDFT1	184420	100	100	100	99.12
FGD1	300546	100	98.24	94.73	84.21
FGF12	601513	100	100	100	99.88
FGF13	300070	100	100	100	97.46
FLNA	300017	99.95	99.05	97.51	95.12
FOLR1	136430	100	100	100	100
FOXG1	164874	99.76	95.53	92.84	88.20
FOXRED1	613622	100	100	100	99.40
FRRS1L	604574	99.23	91.93	85.19	69.41
FXD2	601814	100	100	100	100
GABBR2	607340	97.53	96.67	95.30	89.21
GABRA1	137160	100	100	100	96.96
GABRA2	137140	100	100	100	99.68
GABRA3	305660	100	100	100	97.95
GABRB2	600232	100	100	100	100
GABRB3	137192	100	100	100	100
GABRD	137163	94.27	94.27	92.22	82.81
GABRE	300093	100	99.56	94.26	86.48
GABRG2	137164	100	99.63	96.88	92.07
GAD1	605363	100	100	100	100
GAMT	601240	100	96.09	87.27	77.84
GCSH	238330	100	79.89	72.86	70.19
GLDC	238300	100	100	99.76	97.44
GLRA1	138491	100	100	100	97.81
GLRB	138492	100	99.16	94.17	90.37
GLUD1	138130	100	100	100	97.67
GNAO1	139311	100	100	100	100
GNB1	139380	100	100	100	95.98
GNB2	139390	100	99.75	94.33	80.08
GOSR2	604027	100	100	100	94.62
GPC3	300037	100	100	99.55	95.55
GPHN	603930	97.10	97.10	97.04	92.87
GRIA3	305915	100	100	100	100
GRIK2	138244	100	100	100	97.55
GRIN1	138249	100	100	99.06	93.47
GRIN2A	138253	100	100	100	99.42
GRIN2B	138252	100	100	100	98.83

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GRIN2D	602717	84.48	73.94	66.88	56.11
GRN	138945	100	100	100	97.18
HADH	601609	100	100	100	100
HCFC1	300019	100	100	99.50	95.18
HCN1	602780	100	100	100	99.12
HDAC4	605314	99.52	96.20	94.64	82.86
HID1	605752	100	99.85	97.69	83.66
HLCS	609018	86.97	86.97	86.97	86.62
HNRNPH2	300610	100	100	100	100
HNRNPU	602869	100	98.98	97.17	91.20
HSD17B10	300256	100	100	100	96.57
HSD17B4	601860	100	98.63	96.80	91.72
HUWE1	300697	100	99.65	98.90	93.92
IDH2	147650	100	98.22	91.50	90.70
IER3IP1	609382	100	100	100	100
IFIH1	606951	100	100	100	99.35
INTS8	611351	100	98.64	96.82	88.82
IQSEC2	300522	98.22	95.90	91.53	68.34
IRF2BPL	611720	98.63	96.51	94.91	91.65
JAM3	606871	100	100	100	98.29
KANSL1	612452	100	100	99.30	96.26
KCNA1	176260	100	100	100	100
KCNA2	176262	100	100	100	100
KCNB1	600397	100	100	100	100
KCNC1	176258	100	100	100	100
KCNC2	176256	100	100	100	100
KCND3	605411	100	100	100	98.36
KCNH1	603305	100	100	99.92	98.48
KCNJ10	602208	100	100	100	100
KCNJ11	600937	100	100	100	100
KCNMA1	600150	100	100	94.21	75.47
KCNQ2	602235	100	97.84	92.11	85.20
KCNQ3	602232	98.66	96.71	95.78	92.83
KCNQ5	607357	100	98.26	95.20	91.36
KCNT1	608167	99.93	97.43	92.34	79.58
KCNT2	610044	100	98.88	96.63	95.02
KCTD7	611725	100	100	100	98.73
KDM5C	314690	100	100	99.38	96.36
KIF5A	602821	100	100	99.38	93.97
KIF5C	604593	100	98.78	95.88	87.90

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KMT2A	159555	99.87	99.34	98.67	96.60
KPNA7	614107	100	99.80	97.91	93.91
KPTN	615620	100	100	97.45	84.30
LGI1	604619	100	100	100	100
LIAS	607031	100	100	92.76	92.76
MAST3	612258	99.29	95.96	92.29	83.79
MBD5	611472	100	100	99.65	98.72
MDH2	154100	100	100	100	98.59
MECP2	300005	100	100	100	88.81
MED12	300188	100	99.89	98.40	93.18
MEF2C	600662	100	100	100	95.92
MFSD8	611124	100	100	100	97.84
MLC1	605908	100	100	100	92.60
MOCS1	603707	100	100	97.49	93.02
MOCS2	603708	100	100	100	100
MPDU1	604041	100	100	100	90.01
MTHFR	607093	100	100	99.74	95.92
MTHFS	604197	100	100	100	97.76
MTOR	601231	100	99.77	99.20	96.48
NAPB	611270	100	100	100	100
NBEA	604889	99.38	98.96	98.17	94.46
NDUFA1	300078	100	100	100	100
NDUFA11	612638	100	100	99.93	83.25
NDUFAF1	606934	100	100	100	99.58
NDUFAF2	609653	100	100	97.27	79.30
NDUFAF3	612911	100	100	98.62	90.03
NDUFAF4	611776	100	100	100	92.65
NDUFAF5	612360	99.06	95.12	95.12	91.95
NDUFB3	603839	100	100	100	100
NDUFB9	601445	100	100	100	97.65
NDUFS1	157655	100	100	100	99.40
NDUFS2	602985	100	100	100	93.32
NDUFS3	603846	100	100	100	100
NDUFS4	602694	100	100	100	100
NDUFS6	603848	100	100	100	88.94
NDUFV1	161015	100	100	98.87	93.78
NDUFV2	600532	100	100	99.17	94.16
NECAP1	611623	100	100	100	100
NEDD4L	606384	100	100	99.18	94.15
NEXMIF	300524	100	100	100	100

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NGLY1	610661	100	100	99.43	92.60
NHLRC1	608072	100	100	100	99.83
NPRL2	607072	100	100	100	98.42
NPRL3	600928	100	100	99.77	87.10
NRXN1	600565	100	100	100	100
NSDHL	300275	100	100	100	99.92
NTRK2	600456	100	99.86	98.76	96.66
NUBPL	613621	100	100	100	100
OFD1	300170	100	99.97	99.28	94.92
OGT	300255	100	100	100	99.20
OPHN1	300127	100	100	100	98.90
PACS2	610423	99.47	97.68	94.97	87.75
PAK3	300142	100	100	99.76	93.20
PC	608786	100	100	99.95	96.64
PCDH12	605622	100	100	100	96.48
PCDH19	300460	100	100	100	99.05
PDHA1	300502	100	100	98.88	92.65
PDHB	179060	100	100	100	99.37
PDP1	605993	100	100	100	100
PDX1	600733	100	100	96.79	67.38
PET100	614770	100	100	100	100
PEX1	602136	100	99.82	97.75	93.52
PEX10	602859	100	100	100	94.29
PEX12	601758	100	100	100	100
PEX13	601789	100	100	100	96.20
PEX14	601791	100	100	98.17	83.71
PEX16	603360	100	100	95.63	67.73
PEX19	600279	100	100	95.14	86.96
PEX26	608666	100	100	100	95.81
PEX3	603164	100	100	100	99.12
PEX5	600414	100	100	99.73	93.66
PEX6	601498	100	99.12	95.44	87.85
PGAP1	611655	100	99.36	95.40	86.16
PGAP3	611801	100	100	100	100
PHACTR1	608723	80.56	80.56	78.06	67.22
PHF6	300414	100	100	96.59	73.06
PHGDH	606879	100	100	99.78	94.15
PIGA	311770	100	100	99.88	96.12
PIGN	606097	100	99.91	96.41	82.97
PIGO	614730	100	99.39	97.26	91.81

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PIGS	610271	100	99.69	97.64	91.10
PIGT	610272	100	100	98.63	89.87
PLA2G6	603604	100	100	99.56	94.14
PLCB1	607120	100	100	99.88	95.98
PLP1	300401	100	100	97.94	91.30
PLPBP	604436	100	100	91.78	87.92
PMM2	601785	100	100	100	98.16
PNKP	605610	100	100	98.81	93.63
PNPO	603287	100	100	100	99.30
POLG	174763	100	100	98.60	90.96
PPP2R1A	605983	100	100	98.43	94.29
PPP3CA	114105	100	100	100	99.10
PPT1	600722	100	100	100	96.71
PQBP1	300463	100	100	100	95.57
PRICKLE1	608500	100	100	100	98.63
PRICKLE2	608501	100	100	100	99.66
PRIMA1	613851	88.31	84.04	67.72	50.09
PRRT2	614386	100	100	100	97.93
PSAT1	610936	100	100	100	96.43
PSPH	172480	100	98.52	93.81	76.13
PURA	600473	100	92.85	88.95	87.02
PYCR2	616406	100	100	98.48	87.47
QARS1	603727	100	100	100	97.99
RAB39B	300774	100	100	100	100
RAI1	607642	100	100	99.95	98.22
RANBP2	601181	99.96	98.98	97.95	95.83
RARS2	611524	100	100	100	98.90
RELN	600514	100	100	99.94	98.16
RHOBTB2	607352	100	100	100	98.32
RNASEH2A	606034	100	100	99.81	92.38
RNASEH2B	610326	100	91.35	91.35	91.35
RNASEH2C	610330	100	100	100	100
ROGDI	614574	99.72	95.93	93.99	82.15
RORB	601972	100	100	100	100
RPS6KA3	300075	100	100	99.14	94.66
RRM2B	604712	100	100	100	100
SAMHD1	606754	100	100	100	100
SCAMP5	613766	100	99.76	95.03	70.67
SCARB2	602257	100	100	100	99.01
SCN1A	182389	100	100	100	99.91

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SCN1B	600235	93.06	93.06	93.06	90.97
SCN2A	182390	100	100	99.97	98.32
SCN3A	182391	100	100	99.88	99.25
SCN8A	600702	100	100	99.85	98.35
SEMA6B	608873	99.36	89.71	80.97	70.89
SETBP1	611060	100	100	100	99.94
SHANK3	606230	93.80	89.10	82.66	68.19
SIK1	605705	100	100	100	99.54
SLC12A5	606726	100	99.21	95.99	86.11
SLC13A5	608305	100	100	100	97.99
SLC16A1	600682	100	100	100	100
SLC19A3	606152	99.94	99.94	99.94	99.94
SLC1A3	600111	100	100	100	91.40
SLC25A1	190315	100	100	100	100
SLC25A15	603861	100	100	100	100
SLC25A22	609302	100	100	100	98.43
SLC2A1	138140	100	97.73	97.73	97.73
SLC32A1	616440	100	100	100	97.09
SLC35A2	314375	100	100	95.91	84.43
SLC38A3	604437	100	100	98.95	94.56
SLC4A4	603345	100	100	100	98.80
SLC6A1	137165	99.83	93.69	85.87	80.84
SLC6A5	604159	100	100	100	95.19
SLC6A8	300036	99.70	91.39	77.18	53.90
SLC9A6	300231	100	100	98.84	95.42
SMARCA2	600014	100	100	100	100
SMC1A	300040	100	100	99.90	95.83
SMS	300105	100	100	98.82	96.70
SNAP25	600322	100	100	100	98.28
SNX27	611541	100	100	100	97.45
SON	182465	99.99	99.99	99.60	97.37
SPTAN1	182810	100	100	99.65	96.55
SPTBN1	182790	100	100	99.60	97.26
SRPX2	300642	100	100	98.93	95.05
ST3GAL3	606494	100	100	100	100
ST3GAL5	604402	100	100	100	99.37
STX1B	601485	100	99.77	95.30	89.90
STXBP1	602926	100	100	100	97.49
SUOX	606887	100	100	100	96.61
SYN1	313440	100	97.49	89.24	66.53

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SYNGAP1	603384	99.58	98.03	97.54	92.81
SYNJ1	604297	99.84	97.11	94.83	92.88
SYP	313475	100	94.71	90.70	74.69
SZT2	615463	100	99.59	98.16	91.83
TANGO2	616830	100	100	99.95	96.86
TBC1D24	613577	100	100	99.06	96.31
TBCE	604934	100	100	99.25	96.43
TBCK	616899	100	97.45	96.37	89.67
TCF4	602272	100	99.45	98.49	94.21
TDP2	605764	100	100	100	99.59
THG1L	618802	100	100	100	100
TPP1	607998	100	98.87	96.77	93.28
TREX1	606609	100	100	100	100
TRIM8	606125	100	100	100	99.21
TRIO	601893	98.94	98.12	97.73	95.70
TRPM6	607009	100	99.77	98.46	96.42
TSC1	605284	100	100	100	99.86
TSC2	191092	100	100	99.90	96.43
TSEN54	608755	95.94	95.77	93.88	86.15
TUBA1A	602529	100	100	100	100
TUBB2A	615101	100	98.83	85.30	67.99
TUBG1	191135	100	100	100	100
UBA5	610552	98.76	94.63	91.12	83.09
UBE2A	312180	100	100	100	99.05
UBE3A	601623	100	100	100	98.22
UGDH	603370	100	100	100	99.00
UGP2	191760	100	100	100	99.77
UNC13A	609894	100	99.71	99.01	94.94
UNC13B	605836	65.14	65.14	65.14	64.92
VPS50	616465	99.96	98.48	95.06	89.48
WAC	615049	99.37	97.25	97.25	92.46
WDR26	617424	92.55	87.61	85.58	83.22
WDR45	300526	100	100	100	98.36
WWOX	605131	100	100	100	94.03
XK	314850	100	100	100	98.74
YWHAG	605356	100	100	98.72	89.95
ZDHHC9	300646	100	100	98.94	85.02
ZEB2	605802	100	100	100	100
ZMYND11	608668	100	100	100	100

- OMIM release used: 23-9-2021

HGNC approved	OMIM gene ID (active link to omim.org)	% covered $\geq 10x$	% covered $\geq 20x$	% covered $\geq 30x$	% covered $\geq 50x$
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gene symbol

- The statistics above are based on a set of 104 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