Whole Exome Sequencing
Gene package Autism, version 5.2, 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).

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HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ACADVL	609575	100	99.71	98.00	90.48
ACOX1	609751	100	100	100	98.47
ACSL4	300157	100	100	100	96.52
ADNP	611386	100	100	100	100
ADSL	608222	98.31	98.31	98.31	98.31
AHI1	608894	100	100	100	99.41
ALDH18A1	138250	100	100	100	98.06
ALDH1A3	600463	100	99.78	95.41	85.77
ALDH5A1	610045	100	99.36	94.61	80.34
ALDH7A1	107323	100	100	100	99.34
AMT	238310	100	100	100	97.08
AP1S2	300629	100	100	96.81	83.71
ARID1B	614556	97.28	93.67	88.64	78.80
ARX	300382	84.88	79.12	68.56	50.95
ASH1L	607999	100	100	100	99.35
ASXL3	615115	100	99.86	99.43	98.11
ATP1A2	182340	100	100	99.93	95.75
AUTS2	607270	100	99.61	95.60	80.85
BCKDK	614901	100	100	98.83	93.41

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BRAF	164757	95.04	94.58	93.81	88.27
C12orf57	615140	100	100	100	100
CACNA1C	114205	100	100	99.65	97.43
CAMTA1	611501	98.81	98.81	98.28	93.48
CBS	613381	100	100	100	100
CDKL5	300203	100	99.94	98.62	93.38
CDKN1C	600856	84.56	72.52	60.78	51.01
CHD2	602119	100	99.69	98.24	91.83
CHD7	608892	100	100	99.89	98.69
CHD8	610528	100	100	100	99.48
CLN8	607837	100	100	100	100
CNTNAP2	604569	100	100	99.59	97.84
COL18A1	120328	100	99.84	98.50	87.55
CREBBP	600140	100	99.21	97.19	91.73
CTCF	604167	100	97.46	92.00	84.96
CTNNB1	116806	100	100	100	100
DCHS1	603057	100	100	100	99.67
DCX	300121	100	100	98.63	88.82
DEAF1	602635	96.67	89.74	83.62	77.57
DEPDC5	614191	100	99.98	99.37	97.07
DHCR7	602858	100	100	99.42	94.57
DMD	300377	100	100	99.57	95.52
DMPK	605377	100	100	98.05	86.94
DYRK1A	600855	100	100	100	98.70
EHMT1	607001	99.08	99.04	97.01	92.86
EIF4E	133440	100	100	100	83.02
FGD1	300546	100	98.24	94.73	84.21
FOXG1	164874	99.76	95.53	92.84	88.20
FOXP1	605515	100	100	100	96.07
FTSJ1	300499	100	100	100	99.09
GABRB3	137192	100	100	100	100
GAMT	601240	100	96.09	87.27	77.84
GATM	602360	100	100	100	99.38
GCSH	238330	100	79.89	72.86	70.19
GLDC	238300	100	100	99.76	97.44
GLYCTK	610516	100	100	100	97.11
GNS	607664	100	97.83	96.15	93.60
GRIA3	305915	100	100	100	100
GRIN2A	138253	100	100	100	99.42
GRIN2B	138252	100	100	100	98.83

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H19	103280	No coverage data			
HCN1	602780	100	100	100	99.12
HDC	142704	100	100	100	99.10
HEPACAM	611642	100	98.27	84.11	76.08
HERC2	605837	97.39	95.24	92.48	87.19
HGSNAT	610453	94.18	93.42	91.23	83.05
HOXA1	142955	100	100	100	100
IGF2	147470	100	100	100	96.83
IL1RAPL1	300206	100	100	99.50	96.00
INPP5E	613037	99.81	98.05	95.85	88.74
IQSEC2	300522	98.22	95.90	91.53	68.34
KCNQ1OT1	604115	No coverage data			
KMT5B	610881	100	100	100	97.70
L2HGDH	609584	100	100	99.62	91.41
MAB21L2	604357	100	100	100	99.36
MAGEL2	605283	100	100	99.73	93.03
MAOA	309850	100	100	100	97.95
MAP2K1	176872	100	100	100	97.96
MBD5	611472	100	100	99.65	98.72
MECP2	300005	100	100	100	88.81
MED12	300188	100	99.89	98.40	93.18
MED13	603808	100	99.87	99.47	97.61
MED13L	608771	100	100	99.35	96.30
MEF2C	600662	100	100	100	95.92
METTL23	615262	100	100	100	100
MID1	300552	100	100	100	97.46
MKKS	604896	100	100	100	100
MLC1	605908	100	100	100	92.60
MMUT	609058	100	100	100	100
MYT1L	613084	100	100	99.66	97.37
NAA15	608000	100	100	100	98.02
NAGLU	609701	100	94.32	91.15	85.27
NDP	300658	100	100	100	100
NFIX	164005	100	100	98.22	88.61
NHS	300457	100	98.47	93.63	90.79
NIPBL	608667	100	100	99.65	97.00
NLGN3	300336	100	99.29	98.27	93.57
NLGN4X	300427	100	100	100	100
NOVA2	601991	93.25	86.50	80.59	67.77
NPHP1	607100	99.79	96.88	96.88	95.81

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NR2F1	132890	100	97.29	93.34	85.21
NRXN1	600565	100	100	100	100
NSD1	606681	100	99.87	99.57	97.75
NSUN2	610916	100	97.72	94.39	90.51
OPHN1	300127	100	100	100	98.90
PAFAH1B1	601545	100	100	95.56	93.36
PAH	612349	100	100	100	100
PAX6	607108	100	100	100	100
PCDH19	300460	100	100	100	99.05
PEX7	601757	100	100	99.06	87.85
PHF8	300560	100	100	98.72	92.75
PIGV	610274	100	100	100	100
POGZ	614787	100	100	99.54	96.09
POMGNT1	606822	100	100	100	98.99
POMT1	607423	100	100	99.08	95.04
PQBP1	300463	100	100	100	95.57
PRODH	606810	99.23	96.85	94.69	87.40
PRSS12	606709	100	100	100	97.56
PTCHD1	300828	100	100	99.82	97.67
PTEN	601728	100	100	100	100
RAB39B	300774	100	100	100	100
RAX	601881	100	99.59	90.48	78.83
RPGRIP1L	610937	99.73	98.44	96.86	92.30
RXYLT1	605862	100	100	100	100
SCN2A	182390	100	100	99.97	98.32
SCN8A	600702	100	100	99.85	98.35
SETD2	612778	100	99.64	98.30	96.29
SETD5	615743	100	100	99.85	97.41
SGSH	605270	93.52	93.52	93.52	93.25
SHANK2	603290	97.94	97.84	97.00	92.07
SHANK3	606230	93.80	89.10	82.66	68.19
SLC35A3	605632	100	100	100	100
SLC35C1	605881	100	100	100	99.65
SLC3A1	104614	100	100	100	98.87
SLC6A1	137165	99.83	93.69	85.87	80.84
SLC6A8	300036	99.70	91.39	77.18	53.90
SLC7A9	604144	100	100	100	98.91
SLC9A6	300231	100	100	98.84	95.42
SLC9A9	608396	100	100	100	100
SLITRK1	609678	100	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
SMARCA2	600014	100	100	100	100
SOX10	602229	100	99.66	97.70	90.57
SYNGAP1	603384	99.58	98.03	97.54	92.81
TANC2	615047	100	100	99.68	97.35
TBC1D20	611663	99.05	93.43	93.43	93.43
TBR1	604616	100	100	100	99.72
TCF12	600480	100	100	100	99.58
TM4SF20	615404	100	100	100	100
TRIP12	604506	100	100	99.85	98.05
TSC1	605284	100	100	100	99.86
TSC2	191092	100	100	99.90	96.43
TUBA1A	602529	100	100	100	100
UPF3B	300298	97.99	88.74	80.83	72.83
UQCC2	614461	100	100	100	100
VAMP2	185881	100	100	93.17	92.51
VPS13B	607817	99.28	98.29	97.00	91.79
WAC	615049	99.37	97.25	97.25	92.46
WFS1	606201	100	100	99.11	96.60
XPC	613208	100	100	98.61	88.95
ZBTB20	606025	100	100	99.13	96.55
ZFPM2	603693	100	100	99.70	95.45
ZIC1	600470	100	100	98.79	94.43
ZMYND11	608668	100	100	100	100

- OMIM release used: 18-2-2021
- 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 (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x