Whole Exome Sequencing
Gene package Autism, version 5, 26-2-2021

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/). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy 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 filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). It is not excluded that pathogenic variants 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)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ACADVL	609575	100	99.82	97.92	88.77
ACOX1	609751	100	100	99.87	95.35
ACSL4	300157	100	100	96.89	84.19
ADNP	611386	100	100	100	100
ADSL	608222	98.31	98.31	98.31	98.08
AHI1	608894	100	100	99.93	98.69
ALDH18A1	138250	100	100	99.65	96.64
ALDH1A3	600463	100	98.80	92.73	81.26
ALDH5A1	610045	100	99.09	92.14	79.75
ALDH7A1	107323	100	100	100	95.93
AMT	238310	100	100	100	95.82
AP1S2	300629	100	94.91	87.33	60.78
ARID1B	614556	97.22	93.72	88.85	79.74
ARX	300382	85.61	80.04	71.89	58.82
ASH1L	607999	100	100	99.85	98.58
ASXL3	615115	100	99.60	99.04	95.98
ATP1A2	182340	100	100	99.46	93.95
AUTS2	607270	100	96.55	89.42	63.70
BCKDK	614901	100	100	98.15	92.03

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BRAF	164757	100	99.25	98.19	92.07
C12orf57	615140	100	100	100	100
CACNA1C	114205	100	99.91	99.35	94.84
CAMTA1	611501	98.81	98.61	96.50	90.59
CBS	613381	100	100	100	100
CDKL5	300203	100	97.89	94.24	84.71
CDKN1C	600856	84.21	69.74	58.81	48.73
CHD2	602119	99.87	97.64	94.00	84.58
CHD7	608892	100	99.87	98.61	96.80
CHD8	610528	100	100	99.86	98.43
CLN8	607837	100	100	100	99.67
CNTNAP2	604569	100	100	99.26	95.92
COL18A1	120328	100	99.77	98.50	85.10
CREBBP	600140	99.64	98.59	96.38	88.55
CTCF	604167	99.45	95.46	89.37	82.53
CTNNB1	116806	100	100	100	99.39
DCHS1	603057	100	100	100	98.84
DCX	300121	100	98.56	95.83	82.33
DEAF1	602635	99.15	90.80	83.57	75.87
DEPDC5	614191	100	99.84	98.86	95.68
DHCR7	602858	100	100	98.66	92.36
DMD	300377	100	99.43	96.75	87.68
DMPK	605377	100	99.78	96.78	81.27
DYRK1A	600855	100	100	99.46	96.61
EHMT1	607001	99.08	98.64	96.67	90.52
EIF4E	133440	100	100	95.13	69.30
FGD1	300546	100	98.23	95.76	87.62
FOXG1	164874	98.76	94.08	91.12	86.79
FOXP1	605515	100	100	100	95.29
FTSJ1	300499	100	100	100	97.70
GABRB3	137192	100	100	100	99.25
GAMT	601240	100	95.42	85.45	77.84
GATM	602360	100	100	100	97.45
GCSH	238330	95.40	73.67	72.86	65.51
GLDC	238300	100	100	99.47	95.06
GLYCTK	610516	100	100	100	95.60
GNS	607664	100	97.75	95.87	90.42
GRIA3	305915	100	100	100	90.45
GRIN2A	138253	100	100	100	98.53
GRIN2B	138252	100	100	99.87	98.02

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
H19	103280	No coverage data			
HCN1	602780	100	100	99.86	94.31
HDC	142704	100	100	100	97.55
HEPACAM	611642	100	98.85	82.42	75.76
HERC2	605837	97.43	94.71	91.64	84.39
HGSNAT	610453	94.18	93.19	91.08	81.62
HOXA1	142955	100	100	100	100
IGF2	147470	100	100	100	93.97
IL1RAPL1	300206	100	99.34	97.62	89.23
INPP5E	613037	99.81	97.42	94.23	87.57
IQSEC2	300522	98.35	96.06	91.85	75.50
KCNQ1OT1	604115	No coverage data			
KMT5B	610881	100	100	100	93.68
L2HGDH	609584	100	100	96.57	86.00
MAB21L2	604357	100	100	100	99.18
MAGEL2	605283	100	100	97.36	80.59
MAOA	309850	100	100	99.79	89.85
MAP2K1	176872	100	100	99.21	93.07
MBD5	611472	100	99.86	99.31	97.46
MECP2	300005	100	99.68	97.39	91.06
MED12	300188	100	99.88	97.97	91.23
MED13	603808	100	99.54	98.76	94.55
MED13L	608771	100	99.87	98.57	94.09
MEF2C	600662	100	100	99.24	92.20
METTL23	615262	100	100	100	100
MID1	300552	100	100	100	92.27
MKKS	604896	100	100	100	100
MLC1	605908	100	100	99.07	85.27
MMUT	609058	100	100	100	98.03
MYT1L	613084	100	99.92	99.00	92.55
NAA15	608000	100	100	99.58	90.74
NAGLU	609701	100	94.64	90.80	83.50
NDP	300658	100	100	100	100
NFIX	164005	100	100	97.89	87.43
NHS	300457	100	97.94	94.38	90.30
NIPBL	608667	100	99.77	98.78	95.02
NLGN3	300336	100	99.44	98.25	94.24
NLGN4X	300427	100	100	100	100
NOVA2	601991	92.83	85.54	79.24	61.34
NPHP1	607100	96.88	96.88	96.88	92.57

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NR2F1	132890	100	96.99	93.15	83.18
NRXN1	600565	100	100	100	94.22
NSD1	606681	100	99.68	98.95	96.26
NSUN2	610916	99.89	97.50	93.90	88.08
OPHN1	300127	100	100	99.53	87.77
PAFAH1B1	601545	100	95.52	93.36	92.38
PAH	612349	100	100	100	98.21
PAX6	607108	100	100	100	99.85
PCDH19	300460	100	100	99.85	96.90
PEX7	601757	100	100	96.83	87.17
PHF8	300560	100	99.97	98.72	91.63
PIGV	610274	100	100	100	100
POGZ	614787	100	99.96	98.81	93.86
POMGNT1	606822	100	100	100	98.92
POMT1	607423	100	100	98.66	94.11
PQBP1	300463	100	100	100	95.46
PRODH	606810	99.83	97.31	94.52	85.99
PRSS12	606709	100	100	100	94.21
PTCHD1	300828	100	100	99.43	96.00
PTEN	601728	100	100	100	100
RAB39B	300774	100	100	100	97.20
RAX	601881	100	98.54	88.21	75.09
RPGRIP1L	610937	99.01	97.45	94.84	90.66
RXYLT1	605862	100	100	100	100
SCN2A	182390	100	99.96	99.36	94.90
SCN8A	600702	100	100	99.53	95.79
SETD2	612778	99.99	99.21	97.98	96.10
SETD5	615743	100	100	99.08	94.16
SGSH	605270	93.52	93.52	93.52	92.20
SHANK2	603290	97.94	97.55	96.31	91.43
SHANK3	606230	94.15	88.94	81.69	65.49
SLC35A3	605632	100	100	100	97.83
SLC35C1	605881	100	100	100	100
SLC3A1	104614	100	100	100	97.98
SLC6A1	137165	99.95	94.80	87.24	80.57
SLC6A8	300036	99.54	92.59	81.36	61.87
SLC7A9	604144	100	100	99.76	94.06
SLC9A6	300231	100	98.43	96.80	87.64
SLC9A9	608396	100	100	100	98.43
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	97.99
SOX10	602229	100	98.83	96.64	87.59
SYNGAP1	603384	99.61	98.03	97.03	91.05
TANC2	615047	100	99.97	99.40	95.83
TBC1D20	611663	98.94	93.43	93.43	92.99
TBR1	604616	100	100	100	97.69
TCF12	600480	100	100	100	98.28
TM4SF20	615404	100	100	100	100
TRIP12	604506	100	100	99.44	96.76
TSC1	605284	100	100	100	98.90
TSC2	191092	100	100	99.62	95.03
TUBA1A	602529	100	100	100	100
UPF3B	300298	94.16	83.19	76.18	59.89
UQCC2	614461	100	100	100	97.38
VAMP2	185881	100	100	92.51	90.20
VPS13B	607817	99.15	98.02	95.90	88.67
WAC	615049	98.96	97.25	95.25	89.51
WFS1	606201	100	100	99.40	96.23
XPC	613208	100	99.78	97.93	87.13
ZBTB20	606025	100	100	98.98	96.35
ZFPM2	603693	100	100	98.33	91.93
ZIC1	600470	100	100	99.29	95.43
ZMYND11	608668	100	100	100	97.44

- OMIM release used: 18-2-2021
- The statistics above are based on a set of 100 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