Whole Exome Sequencing
Gene package Autism, version 4, 30-9-2020

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
ACADVL	609575	238	100	100	100
ACOX1	609751	126	100	100	98
ACSL4	300157	47	97	86	72
ADNP	611386	160	100	100	98
ADSL	608222	178	100	100	100
AHI1	608894	65	96	86	77
ALDH18A1	138250	169	100	100	99
ALDH1A3	600463	129	100	100	99
ALDH5A1	610045	121	100	100	98
ALDH7A1	107323	105	100	100	95
AMT	238310	225	100	100	100
AP1S2	300629	33	95	74	53
ARID1B	614556	166	100	100	98
ARX	300382	100	92	89	84
ASH1L	607999	115	100	100	99
ASXL3	615115	119	100	97	94
ATP1A2	182340	189	100	100	100
AUTS2	607270	204	100	99	98
BCKDK	614901	220	100	100	100
BRAF	164757	84	100	99	94
C12orf57	615140	308	100	100	100
CACNA1C	114205	179	100	100	100
CAMTA1	611501	210	100	98	95
CBS	613381	430	100	100	100
CDKL5	300203	74	97	92	86
CDKN1C	600856	156	92	87	83

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
CHD2	602119	82	98	92	84
CHD7	608892	167	100	99	97
CHD8	610528	185	100	100	99
CLN8	607837	195	100	100	100
CNTNAP2	604569	119	100	100	99
COL18A1	120328	247	100	100	100
CREBBP	600140	275	99	99	98
CTCF	604167	114	100	96	92
CTNNB1	116806	133	100	100	99
DCHS1	603057	292	100	100	100
DCX	300121	102	100	96	89
DEAF1	602635	191	100	100	100
DEPDC5	614191	157	100	100	98
DHCR7	602858	243	100	100	100
DMD	300377	68	98	93	87
DMPK	605377	233	100	100	100
DYRK1A	600855	111	100	97	92
EHMT1	607001	176	99	99	98
EIF4E	133440	78	93	71	57
FGD1	300546	126	100	100	100
FOXG1	164874	424	100	97	94
FOXP1	605515	121	100	100	100
FTSJ1	300499	155	100	100	100
GABRB3	137192	160	100	100	100
GAMT	601240	284	100	100	100
GATM	602360	105	100	100	99
GCSH	238330	43	100	85	69
GLDC	238300	118	100	100	98
GLYCTK	610516	298	100	100	100
GNS	607664	105	100	100	99
GRIA3	305915	62	98	91	79
GRIN2A	138253	408	100	100	99
GRIN2B	138252	237	100	100	99
H19	103280	No coverage data			
HCN1	602780	193	100	99	97
HDC	142704	191	100	100	100
HEPACAM	611642	195	100	100	100
HERC2	605837	144	99	94	90
HGSNAT	610453	93	99	94	92
HOXA1	142955	294	100	100	100
IGF2	147470	225	100	100	100
IL1RAPL1	300206	56	100	92	78
INPP5E	613037	259	100	100	100
IQSEC2	300522	128	100	99	98

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
KCNQ1OT1	604115	No coverage data			
KMT5B	610881	86	99	95	89
L2HGDH	609584	73	97	96	93
MAB21L2	604357	511	100	100	100
MAGEL2	605283	355	100	100	100
MAOA	309850	74	100	99	92
MAP2K1	176872	148	100	100	99
MBD5	611472	141	100	99	97
MECP2	300005	200	100	100	99
MED12	300188	99	100	100	98
MED13	603808	93	100	97	91
MED13L	608771	124	100	100	100
MEF2C	600662	157	100	100	99
METTL23	615262	109	100	100	100
MID1	300552	112	100	100	97
MKKS	604896	251	100	100	100
MLC1	605908	181	100	100	95
MMUT	609058	101	100	95	92
MYT1L	613084	174	100	97	91
NAA15	608000	59	99	91	78
NAGLU	609701	182	100	100	100
NDP	300658	155	100	100	100
NFIX	164005	273	100	100	100
NHS	300457	96	100	99	97
NIPBL	608667	93	98	90	82
NLGN3	300336	154	100	100	99
NLGN4X	300427	201	100	100	100
NOVA2	601991	210	99	96	94
NPHP1	607100	89	97	96	89
NR2F1	132890	208	100	99	96
NRXN1	600565	190	100	99	98
NSD1	606681	208	100	99	98
NSUN2	610916	113	100	99	94
OPHN1	300127	71	100	99	91
PAFAH1B1	601545	72	86	86	85
PAH	612349	121	100	100	99
PAX6	607108	267	100	100	100
PCDH19	300460	221	100	100	98
PEX7	601757	94	100	100	94
PHF8	300560	98	100	100	98
PIGV	610274	186	100	100	100
POGZ	614787	154	100	100	99
POMGNT1	606822	228	100	100	100
POMT1	607423	155	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
PQBP1	300463	124	100	100	100
PRODH	606810	222	100	99	98
PRSS12	606709	157	100	100	100
PTCHD1	300828	101	100	100	99
PTEN	601728	100	99	94	87
RAB39B	300774	156	100	100	99
RAX	601881	198	100	100	100
RPGRIP1L	610937	80	95	91	83
RPL5	603634	69	89	85	74
RXYLT1	605862	75	99	91	82
SCN2A	182390	93	98	94	86
SCN8A	600702	137	100	100	98
SETD2	612778	113	100	99	98
SETD5	615743	121	100	99	96
SGSH	605270	272	99	94	94
SHANK2	603290	323	98	98	98
SHANK3	606230	246	99	98	97
SLC35A3	605632	49	93	85	76
SLC35C1	605881	329	100	100	100
SLC3A1	104614	133	99	96	89
SLC6A1	137165	177	100	100	98
SLC6A8	300036	134	100	100	97
SLC7A9	604144	154	100	100	99
SLC9A6	300231	89	98	88	81
SLC9A9	608396	79	100	94	88
SLITRK1	609678	210	100	100	100
SMARCA2	600014	95	100	98	96
SOX10	602229	381	100	100	100
SYNGAP1	603384	201	100	98	98
TANC2	615047	152	100	100	98
TBC1D20	611663	149	100	100	96
TBR1	604616	270	100	100	100
TCF12	600480	94	100	99	96
TM4SF20	615404	75	100	100	99
TRIP12	604506	87	100	99	97
TSC1	605284	153	100	100	100
TSC2	191092	314	100	100	100
TUBA1A	602529	219	100	100	100
UPF3B	300298	40	83	64	48
UQCC2	614461	124	100	100	100
VAMP2	185881	152	100	100	100
VPS13B	607817	98	99	95	89
WAC	615049	78	96	88	82
WFS1	606201	268	100	100	100

HGNC approved gene symbol	OMIM gene ID (active link to omim.org)	median depth	% covered >10x	% covered >20x	% covered >30x
XPC	613208	138	100	100	99
ZBTB20	606025	302	100	100	100
ZFPM2	603693	127	100	100	99
ZIC1	600470	322	100	100	100
ZMYND11	608668	86	100	99	93

- 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