Whole Exome Sequencing
Gene package Autism, version 3.1, 31-1-2020

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	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ACADVL	VLCAD deficiency, 201475	609575	114	100	100	98
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	103	100	100	99
ACSL4	Mental retardation 63, 300387	300157	54	100	98	88
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	75	100	100	100
ADSL	Adenylosuccinase deficiency, 103050	608222	89	100	100	96
AHI1	Joubert syndrome 3, 608629	608894	67	100	98	90
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	80	100	100	98
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	88	100	100	97
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	66	100	97	91
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	69	100	100	91
AMT	Glycine encephalopathy, 605899	238310	130	100	100	100
AP1S2	Mental retardation syndromic 5, 304340	300629	40	100	87	62
ARHGEF6	No OMIM phenotype	300267	47	100	97	83

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ARID1B	Coffin-Siris syndrome 1, 135900	614556	102	100	100	98
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	46	89	79	68
ASH1L	Mental retardation 52, 617796	607999	69	100	100	97
ASXL3	Bainbridge-Ropers syndrome, 615485	615115	69	99	99	97
ATP1A2	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	130	100	100	100
AUTS2	Mental retardation 26, 615834	607270	122	100	100	98
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923	614901	167	100	100	100
BRAF	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706	164757	68	100	100	94
C12orf57	Temtamy syndrome, 218340	615140	141	100	100	100
CACNA1C	Brugada syndrome 3, 611875 Long QT syndrome 8, 618447 Timothy syndrome, 601005	114205	123	100	100	99
CACNA1H	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 Hyperaldosteronism, familial, type IV, 617027	607904	127	100	99	97
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	161	100	99	97
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	127	100	100	100
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	58	100	97	87
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	74	90	83	76
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	72	100	100	96

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CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	87	100	100	97
CHD8	{Autism, susceptibility to, 18}, 615032	610528	85	100	100	98
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	129	100	100	100
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	73	100	100	99
COL18A1	Knobloch syndrome, type 1, 267750	120328	143	100	100	97
CREBBP	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849	600140	85	100	99	94
CTCF	Mental retardation 21, 615502	604167	98	100	100	100
CTNNB1	Colorectal cancer, somatic, 114500 Exudative vitreoretinopathy 7, 617572 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	62	100	100	97
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	128	100	100	100
DCX	Lissencephaly, 300067 Subcortical laminal heterotopia, 300067	300121	57	100	98	92
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation 24, 615828	602635	86	100	97	90
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	83	100	100	97
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	112	100	100	100
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	46	100	96	81
DMPK	Myotonic dystrophy 1, 160900	605377	117	100	100	99
DYRK1A	Mental retardation 7, 614104	600855	75	100	100	98
EHMT1	Kleefstra syndrome 1, 610253	607001	126	99	99	99
EIF4E	{Autism, susceptibility to, 19}, 615091	133440	60	100	96	93

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FGD1	Aarskog-Scott syndrome, 305400 Mental retardation syndromic 16, 305400	300546	71	100	98	95
FOXG1	Rett syndrome, congenital variant, 613454	164874	127	97	90	84
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	80	100	100	98
FTSJ1	Mental retardation 9/44, 309549	300499	91	100	100	93
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	95	100	100	98
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	85	100	99	95
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	65	100	100	98
GCSH	?Glycine encephalopathy, 605899	238330	93	100	89	62
GLDC	Glycine encephalopathy, 605899	238300	67	100	98	91
GLYCTK	D-glyceric aciduria, 220120	610516	136	100	100	100
GNS	Mucopolysaccharidosis type IIID, 252940	607664	68	100	100	98
GRIA3	Mental retardation 94, 300699	305915	51	100	98	88
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	103	100	100	100
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	122	100	100	99
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data			
HCN1	Epileptic encephalopathy, early infantile, 24, 615871 Generalized epilepsy with febrile seizures plus, type 10, 618482	602780	79	100	100	98
HDC	{Gilles de la Tourette syndrome, susceptibility to}, 137580	142704	107	100	100	100
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	97	100	100	99
HERC2	Mental retardation 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	102	100	99	96
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	70	94	94	92
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	126	100	100	100
IGF2	?Growth restriction, severe, with distinctive facies, 616489	147470	100	100	100	100
IL1RAPL1	Mental retardation 21/34, 300143	300206	52	100	98	88

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INPP5E	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	613037	113	100	100	97
IQSEC2	Mental retardation 1/78, 309530	300522	66	98	92	85
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	604115	No coverage data			
KMT5B	Mental retardation 51, 617788	610881	96	100	100	99
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	74	100	100	97
MAB21L2	Microphthalmia/coloboma and skeletal dysplasia syndrome, 615877	604357	170	100	100	100
MAGEL2	Schaaf-Yang syndrome, 615547	605283	88	100	98	93
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	49	100	99	92
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	79	100	100	96
MBD5	Mental retardation 1, 156200	611472	81	100	100	99
MECP2	{Autism susceptibility 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation syndromic, Lubs type, 300260 Mental retardation, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	113	100	100	96
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, 300895 Opitz-Kaveggia syndrome, 305450	300188	68	100	100	97
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	73	100	100	97
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	93	100	99	96
METTL23	Mental retardation 44, 615942	615262	66	100	100	98
MID1	Opitz GBBB syndrome, type I, 300000	300552	89	100	100	93
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	80	100	100	100
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	79	100	100	94
MMUT	Methylmalonic aciduria, mut(0) type, 251000	609058	75	100	100	95
MYT1L	Mental retardation 39, 616521	613084	95	100	100	97
NAA15	Mental retardation 50, 617787	608000	79	100	100	95

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NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	112	100	97	93
NDP	Exudative vitreoretinopathy 2, 305390 Norrie disease, 310600	300658	77	100	100	100
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	154	100	100	100
NHS	Cataract 40, 302200 Nance-Horan syndrome, 302350	300457	57	100	96	90
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	65	100	98	92
NLGN3	{Asperger syndrome susceptibility 1}, 300494 {Autism susceptibility 1}, 300425	300336	100	100	100	98
NLGN4X	{Asperger syndrome susceptibility 2}, 300497 {Autism susceptibility 2}, 300495 Mental retardation, 300495	300427	179	100	100	100
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	58	100	98	88
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	197	100	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	104	100	100	97
NSD1	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	77	100	100	98
NSUN2	Mental retardation 5, 611091	610916	85	100	99	92
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	52	100	96	84
PAFAH1B1	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432	601545	82	100	96	90
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	68	100	99	95

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PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	73	100	100	96
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	113	100	100	97
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	57	100	100	95
PHF8	Mental retardation syndrome, Siderius type, 300263	300560	61	100	98	87
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	96	100	100	100
POGZ	White-Sutton syndrome, 616364	614787	88	100	100	97
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 Retinitis pigmentosa 76, 617123	606822	93	100	100	99
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308	607423	103	100	100	100
PQBP1	Renpenning syndrome, 309500	300463	100	100	100	100
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	98	100	95	86
PRSS12	Mental retardation 1, 249500	606709	94	100	100	99
PTCHD1	{Autism, susceptibility to 4}, 300830	300828	79	100	100	98
PTEN	Cowden syndrome 1, 158350 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 {Meningioma}, 607174 Prostate cancer, somatic, 176807	601728	115	85	78	76
RAB39B	Mental retardation 72, 300271 Waisman syndrome, 311510	300774	59	100	100	100

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RAX	Microphthalmia, isolated 3, 611038	601881	124	100	100	93
RNF135	No OMIM phenotype	611358	83	100	100	99
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	60	98	96	89
RPL5	Diamond-Blackfan anemia 6, 612561	603634	54	99	93	85
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	90	100	100	94
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	95	100	100	97
SCN8A	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 ?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080	600702	110	100	100	98
SETD2	Luscan-Lumish syndrome, 616831	612778	68	100	100	98
SETD5	Mental retardation 23, 615761	615743	85	100	100	98
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	103	100	94	90
SHANK2	{Autism susceptibility 17}, 613436	603290	123	100	99	96
SHANK3	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950	606230	117	99	93	86
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	73	100	99	92
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	146	100	100	100
SLC3A1	Cystinuria, 220100	104614	93	100	100	97
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	115	100	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	103	100	98	95
SLC7A9	Cystinuria, 220100	604144	100	100	100	99
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	64	100	96	84
SLC9A9	{?Autism susceptibility 16}, 613410	608396	62	100	100	96
SLITRK1	Tourette syndrome, 137580 ?Trichotillomania, 613229	609678	89	100	100	100
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	94	98	98	96
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	72	100	97	89
SYNGAP1	Mental retardation 5, 612621	603384	158	98	98	98
TBC1D20	Warburg micro syndrome 4, 615663	611663	74	100	93	93

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TBR1	Intellectual developmental disorder with autism and speech delay, 606053	604616	148	100	100	99
TCF12	Craniosynostosis 3, 615314	600480	72	100	100	98
TM4SF20	{Specific language impairment 5}, 615432	615404	58	100	100	96
TRIP12	Mental retardation 49, 617752	604506	75	100	99	95
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	167	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	184	100	100	100
TUBA1A	Lissencephaly 3, 611603	602529	110	100	100	100
UPF3B	Mental retardation, syndromic 14, 300676	300298	74	100	98	87
UQCC2	Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	127	100	100	100
VPS13B	Cohen syndrome, 216550	607817	75	100	99	96
WAC	Desanto-Shinawi syndrome, 616708	615049	57	100	98	91
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, 614296	606201	196	100	100	100
XPC	Xeroderma pigmentosum, group C, 278720	613208	108	100	100	98
ZBTB20	Primrose syndrome, 259050	606025	149	100	100	100
ZFPM2	Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500 46XY sex reversal 9, 616067	603693	82	100	100	100
ZIC1	Craniosynostosis 6, 616602	600470	255	100	100	100
ZMYND11	Mental retardation 30, 616083	608668	73	100	100	98

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- 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