Whole Exome Sequencing
Gene package Autism, version 2.2, 25-7-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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	81	100	98	93
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	71	100	100	94
ACSL4	Mental retardation, X-linked 63, 300387	300157	51	100	98	87
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	61	100	100	99
ADSL	Adenylosuccinase deficiency, 103050	608222	61	100	99	88
AHI1	Joubert syndrome 3, 608629	608894	51	100	94	73
ALDH18A1	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586	138250	54	100	98	88
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	60	100	98	89
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	42	99	94	71
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	50	100	93	71
AMT	Glycine encephalopathy, 605899	238310	73	100	100	98
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	44	100	80	65
ARHGEF6	Mental retardation, X-linked 46, 300436	300267	38	100	93	70
ARID1B	Coffin-Siris syndrome 1, 135900	614556	67	99	97	90

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ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	42	89	80	65
ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	86	100	100	99
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	91	100	100	100
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	40	100	91	76
AUTS2	Mental retardation, autosomal dominant 26, 615834	607270	60	100	97	91
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923	614901	96	100	100	100
BRAF	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant Nonsmall cell lung cancer Noonan syndrome 7, 613706	164757	53	100	98	84
C12orf57	Temtamy syndrome, 218340	615140	103	100	100	100
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	66	100	99	95
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071	300110	63	100	99	91
CACNA1H	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 Hyperaldosteronism, familial, type IV, 617027	607904	78	100	98	95
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	90	100	98	94
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	42	100	95	69

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CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	70	100	100	96
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	46	100	93	78
CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	56	91	83	74
CEP290	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189	610142	57	100	98	84
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	50	100	98	85
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	60	100	99	93
CHD8	{Autism, susceptibility to, 18}, 615032	610528	64	100	99	92
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	81	100	100	100
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	48	100	99	89
COL18A1	Knobloch syndrome, type 1, 267750	120328	76	100	100	95
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	81	100	99	91
CTCF	Mental retardation, autosomal dominant 21, 615502	604167	70	100	100	99
CTNNB1	Colorectal cancer, somatic, 114500 Hepatocellular carcinoma, somatic, 114550 Medulloblastoma, somatic, 155255 Mental retardation, autosomal dominant 19, 615075 Ovarian cancer, somatic, 167000 Pilomatricoma, somatic, 132600	116806	47	100	100	92
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	94	100	100	100
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067	300121	47	100	96	83
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828	602635	56	99	85	84
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	56	100	98	90
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	84	100	100	100

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DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	38	100	94	71
DMPK	Myotonic dystrophy 1, 160900	605377	69	100	99	93
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	46	100	99	85
DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	54	100	98	87
EHMT1	Kleefstra syndrome, 610253	607001	81	99	99	98
EIF4E	{Autism, susceptibility to, 19}, 615091	133440	51	100	95	86
ELP4	?Aniridia 2, 617141	606985	51	98	89	67
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	60	100	99	95
FOXG1	Rett syndrome, congenital variant, 613454	164874	60	99	91	82
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	56	100	99	89
FOXP2	Speech-language disorder-1, 602081	605317	50	100	98	84
FTSJ1	Mental retardation, X-linked 9/44, 309549	300499	68	100	100	94
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	64	100	99	95
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	60	100	97	90
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	44	100	100	89
GCSH	Glycine encephalopathy, 605899	238330	70	100	91	57
GLDC	Glycine encephalopathy, 605899	238300	50	100	94	82
GLYCTK	D-glyceric aciduria, 220120	610516	108	100	100	100
GNS	Mucopolysaccharidosis type IIID, 252940	607664	46	100	98	88
GRIA3	Mental retardation, X-linked 94, 300699	305915	42	100	97	80
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	76	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970	138252	89	100	100	96
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data	0	0	0
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	80	100	99	93
HDC	{Gilles de la Tourette syndrome, susceptibility to}, 137580	142704	76	100	100	98
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	62	100	100	90

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HERC2	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220	605837	73	100	99	95
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	48	94	94	85
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	104	100	100	100
IGF2	?Growth restriction, severe, with distinctive facies, 616489	613037	73	100	100	100
IL1RAPL1	Mental retardation 21/34, 300143	300206	50	100	99	83
INPP5E	Joubert syndrome 1, 213300; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	300522	80	100	100	96
IQSEC2	Mental retardation 1/78, 309530	604115	56	99	97	88
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	609584	No coverage data	0	0	0
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	47	100	99	89
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	118	100	100	100
MAGEL2	Schaaf-Yang syndrome, 615547	605283	107	100	100	100
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	41	100	96	80
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	55	100	100	91
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	58	100	100	97
MECP2	{Autism susceptibility, X-linked 3}, 300496, Isolated cases, X-linked, Multifactorial Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	88	100	100	94
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450	300188	57	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	56	100	97	87
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	67	100	99	96
METTL23	Mental retardation, autosomal recessive 44, 615942	615262	54	100	100	96
MID1	Opitz GBBB syndrome, type I, 300000	300552	70	100	98	85
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	59	100	100	99
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	49	100	98	86

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MMUT	Methylmalonic aciduria, mut(0) type, 251000	609058	56	100	98	86
MYT1L	Mental retardation, autosomal dominant 39, 616521	613084	67	100	99	94
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	80	100	97	90
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600	300658	57	100	100	100
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	78	100	99	90
NHS	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350	300457	44	100	92	84
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	50	100	96	80
NLGN3	{Asperger syndrome susceptibility, X-linked 1}, 300494, Isolated cases, X-linked, Multifactorial {Autism susceptibility, X-linked 1}, 300425, Isolated cases, X-linked, Multifactorial	300336	66	100	99	94
NLGN4X	{Asperger syndrome susceptibility, X-linked 2}, 300497, Isolated cases, X-linked, Multifactorial {Autism susceptibility, X-linked 2}, 300495, Isolated cases, X-linked, Multifactorial Mental retardation, X-linked, 300495, Isolated cases, X-linked, Multifactorial	300427	153	100	100	100
NLGN4Y	No OMIM phenotype	400028	25	65	59	44
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	39	100	92	67
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	82	100	98	91
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	67	100	99	93
NRXN2	No OMIM phenotype	600566	74	100	99	95
NRXN3	No OMIM phenotype	600567	73	100	100	96
NSD1	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	56	100	99	92
NSUN2	Mental retardation, autosomal recessive 5, 611091	610916	58	100	94	83
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	44	100	92	71
PAFAH1B1	Lissencephaly 1, 607432, Isolated cases Subcortical laminar heterotopia, 607432, Isolated cases	601545	62	100	98	88
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	45	100	97	82

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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	50	100	98	84
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	92	100	99	92
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	43	100	100	87
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263	300560	49	100	96	74
PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	71	100	100	94
POGZ	White-Sutton syndrome, 616364	614787	65	100	98	90
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	63	100	100	95
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	69	100	100	98
PQBP1	Renpenning syndrome, 309500	300463	92	100	100	100
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	60	100	92	81
PRSS12	Mental retardation, autosomal recessive 1, 249500	606709	59	100	100	93
PTCHD1	{Autism, susceptibility to, X-linked 4}, 300830	300828	59	100	100	98

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PTEN	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 {Glioma susceptibility 2}, 613028 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 {Meningioma}, 607174 PTEN hamartoma tumor syndrome {Prostate cancer, somatic}, 176807 Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950	601728	85	92	81	77
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	57	100	96	80
RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510	300774	50	100	100	96
RAI1	Smith-Magenis syndrome, 182290, Isolated cases	607642	99	100	100	100
RAX	Microphthalmia, isolated 3, 611038	601881	72	100	100	89
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	59	100	100	97
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	47	97	94	77
RPL5	Diamond-Blackfan anemia 6, 612561	603634	50	100	96	84
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	58	100	98	78
SATB2	Glass syndrome, 612313	608148	69	100	98	87
SCN1A	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6, 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634	182389	67	100	100	94
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	73	100	99	95
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	72	100	99	91
SETBP1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	70	98	96	94

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SETD2	Luscan-Lumish syndrome, 616831	612778	55	100	99	91
SETD5	Mental retardation, autosomal dominant 23, 615761	615743	57	100	99	90
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	90	100	96	94
SHANK1	No OMIM phenotype	604999	62	98	96	89
SHANK2	{Autism susceptibility 17}, 613436	603290	87	100	98	95
SHANK3	Phelan-McDermid syndrome, 606232, Isolated cases {Schizophrenia 15}, 613950	606230	62	93	78	72
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	55	100	96	76
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	89	100	100	99
SLC3A1	Cystinuria, 220100	104614	63	100	99	91
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	73	100	100	96
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	79	100	97	92
SLC7A9	Cystinuria, 220100	604144	62	100	100	96
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243	300231	54	100	94	82
SLC9A9	?{Autism susceptibility 16}, 613410	608396	42	100	97	82
SLITRK1	Tourette syndrome, 137580 ?Trichotillomania, 613229, Multifactorial	609678	68	100	100	99
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	59	100	100	96
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	61	98	96	85
SMC3	Cornelia de Lange syndrome 3, 610759	606062	56	100	97	83
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	68	100	100	100
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	93	98	98	97
TBC1D20	Warburg micro syndrome 4, 615663	611663	53	98	93	93
TCF12	Craniosynostosis 3, 615314	600480	50	100	99	87
TM4SF20	{Specific language impairment 5}, 615432	615404	42	100	98	86
TRAPPC9	Mental retardation, autosomal recessive 13, 613192	611966	68	100	99	92
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	105	100	100	100
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	104	100	100	100
TUBA1A	Lissencephaly 3, 611603	602529	89	100	100	100

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UBE3A	Angelman syndrome, 105830, Isolated cases	601623	53	100	100	93
UPF3B	Mental retardation, X-linked, syndromic 14, 300676	300298	61	100	97	89
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	80	100	100	89
VPS13B	Cohen syndrome, 216550	607817	54	100	98	87
WAC	Desanto-Shinawi syndrome, 616708	615049	42	100	92	72
WFS1	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome, 222300 Wolfram-like syndrome, autosomal dominant, 614296	606201	106	100	100	100
XPC	Xeroderma pigmentosum, group C, 278720	613208	66	100	98	90
YWHAE	No OMIM phenotype	605066	50	100	99	89
ZBTB20	Primrose syndrome, 259050	606025	123	100	100	100
ZFPM2	Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500 46XY sex reversal 9, 616067	603693	60	100	100	100
ZIC1	Craniosynostosis 6, 616602	600470	110	100	100	100
ZMYND11	Mental retardation, autosomal dominant 30, 616083	608668	53	100	99	90

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "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 96 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 describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 20x