Whole Exome Sequencing
Gene package Autism, version 2, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. 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
ACADVL	VLCAD deficiency, 201475	609575	81	100	97
ACOX1	Peroxisomal acyl-CoA oxidase deficiency, 264470	609751	93	100	100
ACSL4	Mental retardation, X-linked 63, 300387	300157	44	99	91
ADNP	Helsmoortel-van der Aa syndrome, 615873	611386	79	100	99
ADSL	Adenylosuccinase deficiency, 103050	608222	86	100	100
AHI1	Joubert syndrome 3, 608629	608894	77	100	98
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	82	100	99
ALDH1A3	Microphthalmia, isolated 8, 615113	600463	45	98	86
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	97	87
ALDH7A1	Epilepsy, pyridoxine-dependent, 266100	107323	76	100	99
AMT	Glycine encephalopathy, 605899	238310	91	100	100
AP1S2	Mental retardation, X-linked syndromic 5, 304340	300629	32	97	79
ARHGEF6	Mental retardation, X-linked 46, 300436	300267	50	99	88
ARID1B	Coffin-Siris syndrome 1, 135900	614556	83	98	93

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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	24	84	61
ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	109	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	96	100	99
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, 309580	300032	38	98	83
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923	614901	95	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	60	98	88
C12orf57	Temtamy syndrome, 218340	615140	85	100	100
CACNA1C	Brugada syndrome 3, 611875 Timothy syndrome, 601005	114205	96	100	100
CACNA1F	Aland Island eye disease, 300600 Cone-rod dystrophy, X-linked, 3, 300476 Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071	300110	41	99	90
CACNA1H	{Epilepsy, childhood absence, susceptibility to, 6}, 611942 {Epilepsy, idiopathic generalized, susceptibility to, 6}, 611942 Hyperaldosteronism, familial, type IV, 617027	607904	77	98	94
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	100	100	99
CASK	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422	300172	39	98	87
CBS	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200	613381	82	100	99
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94

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CDKN1C	Beckwith-Wiedemann syndrome, 130650 IMAGE syndrome, 614732	600856	46	80	72
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	55	98	92
CHD2	Epileptic encephalopathy, childhood-onset, 615369	602119	75	100	98
CHD7	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370	608892	80	100	98
CHD8	{Autism, susceptibility to, 18}, 615032	610528	83	100	99
CLN8	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003	607837	100	100	100
CNTNAP2	{Autism susceptibility 15}, 612100 Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042	604569	85	100	100
COL18A1	Knobloch syndrome, type 1, 267750	120328	71	100	95
CREBBP	Rubinstein-Taybi syndrome 1, 180849	600140	81	100	97
CTCF	Mental retardation, autosomal dominant 21, 615502	604167	47	96	89
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	81	100	100
DCHS1	Mitral valve prolapse 2, 607829 Van Maldergem syndrome 1, 601390	603057	70	99	97
DCX	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067	300121	47	98	85
DEAF1	?Dyskinesia, seizures, and intellectual developmental disorder, 617171 Mental retardation, autosomal dominant 24, 615828	602635	81	87	82
DEPDC5	Epilepsy, familial focal, with variable foci 1, 604364	614191	53	99	90
DHCR7	Smith-Lemli-Opitz syndrome, 270400	602858	114	100	100
DMD	Becker muscular dystrophy, 300376 Cardiomyopathy, dilated, 3B, 302045 Duchenne muscular dystrophy, 310200	300377	50	99	93
DMPK	Myotonic dystrophy 1, 160900	605377	70	100	97
DPYD	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270	612779	79	98	95

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DYRK1A	Mental retardation, autosomal dominant 7, 614104	600855	99	100	100
EHMT1	Kleefstra syndrome, 610253	607001	97	99	99
EIF4E	{Autism, susceptibility to, 19}, 615091	133440	11	45	17
ELP4	?Aniridia 2, 617141	606985	48	89	78
FGD1	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400	300546	43	99	87
FOXG1	Rett syndrome, congenital variant, 613454	164874	80	83	77
FOXP1	Mental retardation with language impairment and with or without autistic features, 613670	605515	80	100	98
FOXP2	Speech-language disorder-1, 602081	605317	76	100	100
FTSJ1	Mental retardation, X-linked 9/44, 309549	300499	59	100	100
GABRB3	{Epilepsy, childhood absence, susceptibility to, 5}, 612269 Epileptic encephalopathy, early infantile, 43, 617113	137192	94	96	94
GAMT	Cerebral creatine deficiency syndrome 2, 612736	601240	59	99	89
GATM	Cerebral creatine deficiency syndrome 3, 612718	602360	93	100	100
GCSH	Glycine encephalopathy, 605899	238330	14	61	18
GLDC	Glycine encephalopathy, 605899	238300	36	81	62
GLYCTK	D-glyceric aciduria, 220120	610516	92	100	100
GNS	Mucopolysaccharidosis type IIID, 252940	607664	84	100	93
GRIA3	Mental retardation, X-linked 94, 300699	305915	54	100	98
GRIN2A	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570	138253	80	100	100
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970	138252	95	100	100
H19	Beckwith-Wiedemann syndrome, 130650 Silver-Russell syndrome, 180860 Wilms tumor 2, 194071	103280	No coverage data		
HCN1	Epileptic encephalopathy, early infantile, 24, 615871	602780	70	100	99
HDC	{Gilles de la Tourette syndrome, susceptibility to}, 137580	142704	98	100	100
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926	611642	63	99	83
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	37	73	63
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 Retinitis pigmentosa 73, 616544	610453	69	94	91
HOXA1	Athabaskan brainstem dysgenesis syndrome, 601536 Bosley-Salih-Alorainy syndrome, 601536	142955	108	100	100
IGF2	?Growth restriction, severe, with distinctive facies, 616489	613037	72	100	100
IL1RAPL1	Mental retardation 21/34, 300143	300206	50	100	100

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INPP5E	Joubert syndrome 1, 213300; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156	300522	71	98	92
IQSEC2	Mental retardation 1/78, 309530	604115	38	94	84
KCNQ1OT1	Beckwith-Wiedemann syndrome, 130650	609584	No coverage data		
AUTS2	Mental retardation, autosomal dominant 26, 615834	607270	81	99	93
L2HGDH	L-2-hydroxyglutaric aciduria, 236792	609584	64	100	92
MAB21L2	Microphthalmia, syndromic 14, 615877	604357	96	100	100
MAGEL2	Schaaf-Yang syndrome, 615547	605283	70	100	97
MAOA	{Antisocial behavior}, 300615 Brunner syndrome, 300615	309850	39	100	86
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	176872	87	100	100
MBD5	Mental retardation, autosomal dominant 1, 156200	611472	82	100	100
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	52	99	81
MED12	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450	300188	47	100	91
MED13L	Mental retardation and distinctive facial features with or without cardiac defects, 616789 Transposition of the great arteries, dextro-looped 1, 608808	608771	42	100	100
MEF2C	Chromosome 5q14.3 deletion syndrome, 613443 (4) Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443	600662	66	100	98
METTL23	Mental retardation, autosomal recessive 44, 615942	615262	77	100	99
MID1	Opitz GBBB syndrome, type I, 300000	300552	57	100	99
MKKS	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700	604896	106	100	100
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	71	100	96
MUT	Methylmalonic aciduria, mut(0) type, 251000	609058	81	100	100
MYT1L	Mental retardation, autosomal dominant 39, 616521	613084	89	100	99
NAGLU	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920	609701	81	100	93
NDP	Exudative vitreoretinopathy 2, X-linked, 305390 Norrie disease, 310600	300658	68	100	100
NFIX	Marshall-Smith syndrome, 602535 Sotos syndrome 2, 614753	164005	90	100	100

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NHS	Cataract 40, X-linked, 302200 Nance-Horan syndrome, 302350	300457	48	95	92
NIPBL	Cornelia de Lange syndrome 1, 122470	608667	64	98	93
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	49	100	97
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	74	100	100
NLGN4Y	No OMIM phenotype	400028	54	98	94
NPHP1	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900	607100	78	100	99
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722	132890	123	100	100
NRXN1	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332	600565	102	100	99
NRXN2	No OMIM phenotype	600566	74	97	91
NRXN3	No OMIM phenotype	600567	84	100	99
NSD1	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550	606681	90	100	99
NSUN2	Mental retardation, autosomal recessive 5, 611091	610916	78	100	95
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	42	100	97
PAFAH1B1	Lissencephaly 1, 607432, Isolated cases Subcortical laminar heterotopia, 607432, Isolated cases	601545	68	100	97
PAH	[Hyperphenylalaninemia, non-PKU mild], 261600 Phenylketonuria, 261600	612349	112	100	100
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	62	100	99
PCDH19	Epileptic encephalopathy, early infantile, 9, 300088	300460	57	100	97
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	68	100	89
PHF8	Mental retardation syndrome, X-linked, Siderius type, 300263	300560	52	99	90

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PIGV	Hyperphosphatasia with mental retardation syndrome 1, 239300	610274	102	100	100
POGZ	White-Sutton syndrome, 616364	614787	82	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	82	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	90	100	100
PQBP1	Renpenning syndrome, 309500	300463	62	100	100
PRODH	Hyperprolinemia, type I, 239500 {Schizophrenia, susceptibility to, 4}, 600850	606810	51	95	83
PRSS12	Mental retardation, autosomal recessive 1, 249500	606709	82	100	98
PTCHD1	{Autism, susceptibility to, X-linked 4}, 300830	300828	54	100	99
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	57	77	73
PTPN11	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950	176876	68	99	99
RAB39B	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510	300774	48	100	97
RAI1	Smith-Magenis syndrome, 182290, Isolated cases	607642	91	100	100
RAX	Microphthalmia, isolated 3, 611038	601881	77	96	80
RNF135	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192	611358	57	100	97
RPGRIP1L	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561	610937	77	97	95
RPL5	Diamond-Blackfan anemia 6, 612561	603634	22	64	40

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SATB2	Glass syndrome, 612313	608148	67	100	99
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	100	100	99
SCN2A	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745	182390	100	100	99
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	97	100	99
SETBP1	Mental retardation, autosomal dominant 29, 616078 Schinzel-Giedion midface retraction syndrome, 269150	611060	74	98	97
SETD2	Luscan-Lumish syndrome, 616831	612778	75	100	98
SETD5	Mental retardation, autosomal dominant 23, 615761	615743	57	99	96
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900	605270	86	94	94
SHANK1	No OMIM phenotype	604999	39	91	73
SHANK2	{Autism susceptibility 17}, 613436	603290	81	100	100
SHANK3	Phelan-McDermid syndrome, 606232, Isolated cases {Schizophrenia 15}, 613950	606230	56	93	82
SLC35A3	?Arthrogryposis, mental retardation, and seizures, 615553	605632	30	94	67
SLC35C1	Congenital disorder of glycosylation, type IIc, 266265	605881	103	100	100
SLC3A1	Cystinuria, 220100	104614	99	100	100
SLC6A1	Myoclonic-atonic epilepsy, 616421	137165	86	100	100
SLC6A8	Cerebral creatine deficiency syndrome 1, 300352	300036	19	81	42
SLC7A9	Cystinuria, 220100	604144	98	100	100
SLC9A6	Mental retardation, X-linked syndromic, Christianson type, 300243	300231	50	100	96
SLC9A9	?{Autism susceptibility 16}, 613410	608396	68	100	98
SLITRK1	Tourette syndrome, 137580 ?Trichotillomania, 613229, Multifactorial	609678	62	100	100
SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900	600993	76	100	98
SMARCA2	Nicolaides-Baraitser syndrome, 601358	600014	78	98	96
SMC3	Cornelia de Lange syndrome 3, 610759	606062	50	93	84
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	45	99	87
SYNGAP1	Mental retardation, autosomal dominant 5, 612621	603384	55	95	82

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TBC1D20	Warburg micro syndrome 4, 615663	611663	51	93	85
TCF12	Craniosynostosis 3, 615314	600480	51	100	94
TM4SF20	{Specific language impairment 5}, 615432	615404	61	100	100
TMEM5	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041	605862	106	100	99
TRAPPC9	Mental retardation, autosomal recessive 13, 613192	611966	85	100	99
TSC1	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100	605284	94	100	99
TSC2	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254	191092	94	100	100
TUBA1A	Lissencephaly 3, 611603	602529	34	93	78
UBE3A	Angelman syndrome, 105830, Isolated cases	601623	61	100	99
UPF3B	Mental retardation, X-linked, syndromic 14, 300676	300298	32	99	83
UQCC2	?Mitochondrial complex III deficiency, nuclear type 7, 615824	614461	51	100	91
VPS13B	Cohen syndrome, 216550	607817	83	100	99
WAC	Desanto-Shinawi syndrome, 616708	615049	48	97	84
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	102	100	100
XPC	Xeroderma pigmentosum, group C, 278720	613208	95	100	100
YWHAE	No OMIM phenotype	605066	56	99	80
ZBTB20	Primrose syndrome, 259050	606025	91	100	99
ZFPM2	Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500 46XY sex reversal 9, 616067	603693	78	100	100
ZIC1	Craniosynostosis 6, 616602	600470	108	100	100
ZMYND11	Mental retardation, autosomal dominant 30, 616083	608668	43	98	88

- 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 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x

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