

## Whole Exome Sequencing

### Gene package Hearing impairment, version 5, 30-7-2018



#### 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).



**Dept. Clinical Genetics**

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	208	100	100	100
ACTG1	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717	102560	174	100	100	100
ADCY1	?Deafness, autosomal recessive 44, 610154	103072	101	97	96	93
ADGRV1	?Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472	602851	67	100	99	93
AIFM1	Combined oxidative phosphorylation deficiency 6, 300816 Cowchock syndrome, 310490 Deafness, X-linked 5, 300614	300169	45	100	95	78
APOPT1	Mitochondrial complex IV deficiency, 220110	616003	64	100	100	93
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	134	100	100	100
ATP2B2	{Deafness, autosomal recessive 12, modifier of}, 601386	108733	128	100	100	99
ATP6V1B1	Renal tubular acidosis with deafness, 267300	192132	120	100	100	100

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BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	165	100	100	100
BDP1	No OMIM phenotype	607012	49	99	92	77
BSND	Bartter syndrome, type 4a, 602522 Sensorineural deafness with mild renal dysfunction, 602522	606412	114	100	100	100
CABP2	Deafness, autosomal recessive 93, 614899	607314	81	100	100	95
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities, 615474 Sinoatrial node dysfunction and deafness, 614896	114206	90	100	99	96
CCDC50	?Deafness, autosomal dominant 44, 607453	611051	62	100	100	94
CD164	?Deafness, autosomal dominant 66, 616969	603356	71	100	100	92
CDC14A	Deafness, autosomal recessive 105, 616958	603504	67	100	99	89
CDH23	Deafness, autosomal recessive 12, 601386 {Pituitary adenoma 5, multiple types}, 617540 Usher syndrome, type 1D, 601067 Usher syndrome, type 1D/F digenic, 601067	605516	123	100	100	100
CEACAM16	Deafness, autosomal dominant 4B, 614614	614591	84	100	100	100
CEP78	Cone-rod dystrophy and hearing loss, 617236	617110	65	100	98	85
CIB2	Deafness, autosomal recessive 48, 609439 Usher syndrome, type II, 614869	605564	175	100	100	100
CLDN14	Deafness, autosomal recessive 29, 614035	605608	59	100	94	87
CLIC5	?Deafness, autosomal recessive 103, 616042	607293	65	100	100	99
CLPP	Perrault syndrome 3, 614129	601119	120	100	100	100
CLRN1	Retinitis pigmentosa 61, 614180 Usher syndrome, type 3A, 276902	606397	77	100	100	93
COCH	Deafness, autosomal dominant 9, 601369	603196	60	98	95	92
COL11A1	Fibrochondrogenesis 1, 228520 {Lumbar disc herniation, susceptibility to}, 603932 Marshall syndrome, 154780 Stickler syndrome, type II, 604841	120280	53	100	98	84
COL11A2	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegapiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegapiphyseal dysplasia, autosomal recessive, 215150	120290	96	100	100	98

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COL2A1	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler syndrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia	120140	92	100	100	98
COL4A3	Alport syndrome, autosomal dominant, 104200 Alport syndrome, autosomal recessive, 203780 Hematuria, benign familial, 141200	120070	54	100	95	83
COL4A4	Alport syndrome, autosomal recessive, 203780 Hematuria, familial benign	120131	61	100	99	91
COL4A5	Alport syndrome, 301050	303630	35	99	84	54
COL4A6	?Deafness, X-linked 6, 300914	303631	53	100	97	82
COL9A1	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134	120210	57	100	97	87
COL9A2	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284	120260	99	100	100	96
CRYM	Deafness, autosomal dominant 40, 616357	123740	66	100	100	97
DCDC2	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394	605755	86	100	99	93
DIABLO	Deafness, autosomal dominant 64, 614152	605219	96	100	100	98
DIAPH1	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632	602121	77	100	98	88
DIAPH3	Auditory neuropathy, autosomal dominant, 1, 609129	614567	55	100	98	85
DMXL2	?Deafness, autosomal dominant 71, 617605 ?Polyendocrine-polyneuropathy syndrome, 616113	612186	58	100	98	89

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DSPP	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500	125485	31	78	56	46
EDN3	Central hypoventilation syndrome, congenital, 209880 {Hirschsprung disease, susceptibility to, 4}, 613712 Waardenburg syndrome, type 4B, 613265	131242	85	100	100	100
EDNRB	ABCD syndrome, 600501 {Hirschsprung disease, susceptibility to, 2}, 600155 Waardenburg syndrome, type 4A, 277580	131244	86	100	100	99
ELMOD3	?Deafness, autosomal recessive 88, 615429	615427	88	100	100	94
EPS8	?Deafness, autosomal recessive 102, 615974	600206	59	100	99	89
EPS8L2	Deafness autosomal recessive 106, 617637	614988	120	100	98	94
ERAL1	Perrault syndrome 6, 617565	607435	111	100	100	100
ESPN	Deafness, autosomal recessive 36, 609006 Deafness, neurosensory, without vestibular involvement, autosomal dominant	606351	101	98	89	81
ESRP1	?Deafness, autosomal recessive 109, 618013	612959	56	100	98	88
ESRRB	Deafness, autosomal recessive 35, 608565	602167	143	100	100	100
EXOSC2	Short stature, hearing loss, retinitis pigmentosa, and distinctive facies, 617763	602238	61	100	100	95
EYA1	Anterior segment anomalies with or without cataract, 602588 Branchiotoic syndrome 1, 602588 Branchiotoic syndrome 1, with or without cataracts, 113650 ?Otofaciocervical syndrome, 166780	601653	80	100	100	92
EYA4	?Cardiomyopathy, dilated, 1J, 605362 Deafness, autosomal dominant 10, 601316	603550	53	100	99	88
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706	164950	118	100	100	100
FOXI1	Enlarged vestibular aqueduct, 600791	601093	135	100	100	100
GAB1	?Deafness, autosomal recessive 26, 605428	604439	63	100	99	95
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255	131320	158	100	100	100
GIPC3	Deafness, autosomal recessive 15, 601869	608792	107	99	93	91
GJB2	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500	121011	140	100	100	100

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GJB3	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratoderma variabilis et progressiva 1, 133200	603324	156	100	100	100
GJB6	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500	604418	65	100	100	96
GPSM2	Chudley-McCullough syndrome, 604213	609245	80	100	99	93
GRHL2	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029	608576	69	100	100	96
GRXCR1	Deafness, autosomal recessive 25, 613285	613283	80	100	100	97
GRXCR2	?Deafness, autosomal recessive 101, 615837	615762	97	100	100	100
GSDME	Deafness, autosomal dominant 5, 600994	608798	91	100	100	99
HARS	Charcot-Marie-Tooth disease, axonal, type 2W, 616625 Usher syndrome type 3B, 614504	142810	90	100	100	98
HARS2	?Perrault syndrome 2, 614926	600783	66	100	100	97
HGF	Deafness, autosomal recessive 39, 608265	142409	61	100	99	93
HOMER2	?Deafness, autosomal dominant 68, 616707	604799	114	99	98	98
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	54	100	96	87
IFNLR1	No OMIM phenotype	607404	84	100	95	94
ILDR1	Deafness, autosomal recessive 42, 609646	609739	89	100	98	93
KARS	?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 Deafness, autosomal recessive 89, 613916	601421	117	100	100	95
KCNE1	Jervell and Lange-Nielsen syndrome 2, 612347 Long QT syndrome 5, 613695	176261	234	100	100	100
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	155	100	100	100
KCNQ1	Atrial fibrillation, familial, 3, 607554 Jervell and Lange-Nielsen syndrome, 220400 Long QT syndrome 1, 192500 {Long QT syndrome 1, acquired, susceptibility to}, 192500 Short QT syndrome 2, 609621	607542	155	96	93	92
KCNQ4	Deafness, autosomal dominant 2A, 600101	603537	139	98	93	89

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KITLG	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664	184745	72	100	100	91
LARS2	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300	604544	68	100	100	99
LHFPL5	Deafness, autosomal recessive 67, 610265	609427	156	100	100	100
LMX1A	No OMIM phenotype	600298	72	100	100	98
LOXHD1	Deafness, autosomal recessive 77, 613079	613072	106	100	100	98
LRTOMT	Deafness, autosomal recessive 63, 611451	612414	99	100	100	100
MARVELD2	Deafness, autosomal recessive 49, 610153	610572	76	100	100	96
MCM2	?Deafness, autosomal dominant 70, 616968	116945	112	100	100	99
MET	?Deafness, autosomal recessive 97, 616705 Hepatocellular carcinoma, childhood type, somatic, 114550 {Osteofibrous dysplasia, susceptibility to}, 607278 Renal cell carcinoma, papillary, 1, familial and somatic, 605074	164860	55	100	99	91
MIR96	Deafness, autosomal dominant 50, 613074	611606	No coverage data			
MITF	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470	156845	72	100	100	97
MPZL2	No OMIM phenotype	604873	54	100	100	94
MSRB3	Deafness, autosomal recessive 74, 613718	613719	62	100	98	89
MYH14	Deafness, autosomal dominant 4A, 600652 ?Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369	608568	112	100	100	98
MYH9	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100	160775	121	100	100	99
MYO15A	Deafness, autosomal recessive 3, 600316	602666	122	100	99	97
MYO3A	Deafness, autosomal recessive 30, 607101	606808	62	100	98	90
MYO6	Deafness, autosomal dominant 22, 606346 Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 Deafness, autosomal recessive 37, 607821	600970	71	100	98	89
MYO7A	Deafness, autosomal dominant 11, 601317 Deafness, autosomal recessive 2, 600060 Usher syndrome, type 1B, 276900	276903	105	100	100	99
NARS2	Combined oxidative phosphorylation deficiency 24, 616239	612803	47	99	96	84

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NLRP3	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900	606416	108	100	100	100
OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	53	100	96	83
OSBPL2	Deafness, autosomal dominant 67, 616340	606731	95	100	100	99
OTOA	Deafness, autosomal recessive 22, 607039	607038	92	100	100	96
OTOF	Auditory neuropathy, autosomal recessive, 1, 601071 Deafness, autosomal recessive 9, 601071	603681	107	100	100	100
OTOG	Deafness, autosomal recessive 18B, 614945	604487	112	100	100	99
OTOGL	Deafness, autosomal recessive 84B, 614944	614925	65	100	98	89
P2RX2	Deafness, autosomal dominant 41, 608224	600844	119	100	100	100
PAX3	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820	606597	94	100	100	100
PCDH15	Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067 Usher syndrome, type 1F, 602083	605514	60	100	98	89
PDE1C	No OMIM phenotype	602987	55	100	96	86
PDZD7	Deafness, autosomal recessive 57, 618003 {Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472	612971	82	100	100	98
PET100	Mitochondrial complex IV deficiency, 220110	614770	70	99	66	66
PEX1	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539	602136	52	100	98	90
PEX6	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863	601498	97	100	100	96
PJKV	Deafness, autosomal recessive 59, 610220	610219	82	100	100	96
PNPT1	Combined oxidative phosphorylation deficiency 13, 614932 Deafness, autosomal recessive 70, 614934	610316	51	100	95	74

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POU3F4	Deafness, X-linked 2, 304400	300039	97	100	100	100
POU4F3	Deafness, autosomal dominant 15, 602459	602460	205	100	100	100
PPIP5K2	No OMIM phenotype	611648	40	99	90	70
PRKCB	No OMIM phenotype	176970	64	100	100	98
PRPS1	Arts syndrome, 301835 Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 Deafness, X-linked 1, 304500, X-linked Gout, PRPS-related, 300661 Phosphoribosylpyrophosphate synthetase superactivity, 300661	311850	44	100	96	81
PTPRQ	Deafness, autosomal dominant 73, 617663 Deafness, autosomal recessive 84A, 613391	603317	69	100	98	91
RAI1	Smith-Magenis syndrome, 182290	607642	167	100	100	100
RDX	Deafness, autosomal recessive 24, 611022	179410	47	100	89	72
RIPOR2	?Deafness, autosomal recessive 104, 616515	611410	67	100	100	95
ROR1	?Deafness, autosomal recessive 108, 617654	602336	72	98	96	95
S1PR2	Deafness, autosomal recessive 68, 610419	605111	142	100	100	100
SERPINB6	?Deafness, autosomal recessive 91, 613453	173321	82	100	100	94
SIX1	Branchiotoxic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192	601205	115	100	100	99
SIX5	Branchiotoorenal syndrome 2, 610896	600963	76	100	96	88
SLC17A8	Deafness, autosomal dominant 25, 605583	607557	56	100	100	96
SLC22A4	{Rheumatoid arthritis, susceptibility to}, 180300	604190	89	100	99	94
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 Pendred syndrome, 274600	605646	60	100	99	87
SLC26A5	?Deafness, autosomal recessive 61, 613865	604943	51	100	99	89
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539	603690	60	100	96	81
SLC44A4	?Deafness, autosomal dominant 72, 617606	606107	83	100	100	99
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	166	100	100	100
SLITRK6	Deafness and myopia, 221200	609681	62	100	100	97
SMPX	Deafness, X-linked 4, 300066	300226	43	100	100	75
SNAI2	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890	602150	56	100	100	100
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	67	100	96	86
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome, 616577	613940	59	100	99	92
STRC	Deafness, autosomal recessive 16, 603720	606440	158	100	100	99



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SYNE4	Deafness, autosomal recessive 76, 615540	615535	107	100	100	100
TBC1D24	DOORS syndrome, 220500 Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021	613577	147	100	100	99
TECTA	Deafness, autosomal dominant 8/12, 601543 Deafness, autosomal recessive 21, 603629	602574	115	100	100	97
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	131	100	100	100
TJP2	Cholestasis, progressive familial intrahepatic 4, 615878 Hypercholanemia, familial, 607748	607709	75	100	100	97
TMC1	Deafness, autosomal dominant 36, 606705 Deafness, autosomal recessive 7, 600974	606706	51	100	98	88
TMEM132E	No OMIM phenotype	616178	110	100	99	96
TMIE	Deafness, autosomal recessive 6, 600971	607237	77	100	99	99
TMPRSS3	Deafness, autosomal recessive 8/10, 601072	605511	67	100	100	96
TMTC2	No OMIM phenotype	615856	68	100	99	96
TNC	Deafness, autosomal dominant 56, 615629	187380	98	100	99	97
TPRN	Deafness, autosomal recessive 79, 613307	613354	100	91	85	78
TRIOBP	Deafness, autosomal recessive 28, 609823	609761	143	100	99	97
TSPEAR	?Deafness, autosomal recessive 98, 614861	612920	102	100	100	100
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286	606075	130	100	100	100
TYR	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 Waardenburg syndrome/albinism, digenic, 103470	606933	77	100	100	96
USH1C	Deafness, autosomal recessive 18A, 602092 Usher syndrome, type 1C, 276904	605242	71	100	95	86
USH1G	Usher syndrome, type 1G, 606943	607696	181	100	100	100
USH2A	Retinitis pigmentosa 39, 613809 Usher syndrome, type 2A, 276901	608400	70	100	99	96
WBP2	Deafness, autosomal recessive 107, 617639	606962	58	100	96	88

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WFS1	?Cataract 41, 116400 Deafness, autosomal dominant 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, autosomal dominant, 614296	606201	175	100	100	100
WHRN	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383	607928	113	100	100	99
YAP1	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433	606608	63	100	99	87

- Gene symbols according HGNC
- OMIM release used: 4-7-2018
- "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 95 samples
- Median depth is the median of the mean sequence depth over the protein coding exons ( $\pm 10$ bp flanking introns) of the longest transcript
- % Covered 10x describes the percentage of a gene's coding sequence ( $\pm 10$ bp flanking introns) that is covered at least 10x
- % Covered 20x describes the percentage of a gene's coding sequence ( $\pm 10$ bp flanking introns) that is covered at least 20x
- % Covered 30x describes the percentage of a gene's coding sequence ( $\pm 10$ bp flanking introns) that is covered at least 30x