UNIVERSITY OF IOWA HEALTH CARE

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OtoSCOPE® GENETIC TESTING

Version 9 (224 genes)

Gene	Hearing Loss Phenotypes	OMIM Gene ID	Inheritance
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (PHARC syndrome)	613599	AR
ACTB	Baraitser-Winter syndrome 1	102630	AD
ACTG1	Deafness, autosomal dominant 20/Deafness, autosomal dominant 26/Baraitser-Winter syndrome 2	102560	AD
ADCY1	Deafness, autosomal recessive 44	103072	AR
ADGRV1	Usher syndrome type 2C	602851	AR
AIFM1	Auditory neuropathy, x-linked 1/Deafness, X-linked 5	300169	XLR
ALMS1	Alström syndrome	606844	AR
AMMECR1	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis	300195	XLR
ANKH	Craniometaphyseal dysplasia	605145	AD
ATP2B2	Deafness, autosomal dominant*	108733	AD
ATP6V0A4	Renal tubular acidosis, distal, 3, with or without sensorineural hearing loss	605239	AR
ATP6V1B1	Renal tubular acidosis, distal, 2, with progressive sensorineural hearing loss	192132	AR
ATP6V1B2	Deafness, Congenital, with Onychodystrophy, autosomal dominant (DDOD syndrome)	606939	AD
BCS1L	Bjornstad syndrome	603647	AR
BDP1	Deafness, autosomal recessive 112	607012	AR
BSND	Deafness, autosomal recessive 73/Bartter syndrome type 4A	606412	AR
BTD	Biotinidase deficiency	609019	AR
CABP2	Deafness, autosomal recessive 93	607314	AR
CACNA1D	Sinoatrial node dysfunction and deafness (SANDD)	114206	AR
CCDC50	Deafness, autosomal dominant 44	611051	AD
CD164	Deafness, autosomal dominant 66	603356	AD
CDC14A	Deafness, autosomal recessive 105/Deafness, autosomal recessive 32/Hearing impairment infertile male syndrome	603504	AR
CDH23	Deafness, autosomal recessive 12/Usher syndrome type 1D	605516	AR
CEACAM16	Deafness, autosomal dominant 4B/Deafness, autosomal recessive 113	614591	AD/AR
CEP78	Cone-rod dystrophy and hearing loss 1	617110	AR
CHD7	CHARGE syndrome	608892	AD
CHSY1	Temtamy preaxial brachydactyly syndrome	608183	AR
CIB2	Deafness, autosomal recessive 48	605564	AR
CISD2	Wolfram syndrome 2	611507	AR
CLDN14	Deafness, autosomal recessive 29	605608	AR
CLDN9	Deafness, autosomal recessive*	615799	AR
CLIC5	Deafness, autosomal recessive 103	607293	AR
CLPP	Perrault syndrome type 3	601119	AR
CLRN1	Usher syndrome type 3A	606397	AR
COCH	Deafness, autosomal dominant 9/Deafness, autosomal recessive 110	603196	AD/AR
COL11A1	Deafness, autosomal dominant 37/Stickler syndrome type 2/Marshall syndrome	120280	AD
COL11A2	Deafness, autosomal recessive 53/Deafness, autosomal dominant 13/Stickler syndrome type 3/Otospondylomegaepiphyseal dysplasia, autosomal dominant/Otospondylomegaepiphyseal dysplasia, autosomal recessive	120290	AD/AR
COL2A1	Stickler syndrome 1	120140	AD
COL4A3	Alport syndrome 2, autosomal recessive/Alport syndrome 3, autosomal dominant	120070	AD/AR
COL4A4	Alport syndrome 2, autosomal recessive	120131	AR
COL4A5	Alport syndrome 1, X-linked	303630	XLD
COL4A6	Deafness, X-linked 6	303631	XLR
COL9A1	Stickler syndrome 4	120210	AR
COL9A2	Stickler syndrome 5	120260	AR
COL9A3	Stickler syndrome	120260	AR
CRYM	Deafness, autosomal dominant 40	123740	AD
DCAF17	Woodhouse-Sakati syndrome	612515	AR
DCDC2	Deafness, autosomal recessive 66	605755	AR
DIABLO	Deafness, autosomal dominant 64	605219	AD
DIAPH1	Deafness, autosomal dominant 1, with or without thrombocytopenia	602121	AD
DIAPH3	Auditory neuropathy, autosomal dominant, 1	614567	AD
DLX5	Split-hand/foot malformation 1 with sensorineural hearing loss	600028	AD/AR
DMXL2	Deafness, autosomal dominant 71	612186	AD
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant/Neuropathy, hereditary sensory, type 1E *(DNMT1* -methylopathies)	126375	AD
DSPP	Deafness, autosomal dominant 39 with dentinogenesis imperfecta	125485	AD

EDN3	Waardenburg syndrome type 4B	131242	AD/AR
EDNRB	Waardenburg syndrome type 4A	131244	AD/AR
ELMOD3	Deafness, autosomal recessive 88	615427	AR
EPS8	Deafness, autosomal recessive 102	600206	AR
EPS8L2	Deafness, autosomal recessive 106	614988	AR
ERAL1	Perrault syndrome type 6	607435	AR
ESPN	Deafness, autosomal recessive 36	606351	AR
ESRRB	Deafness, autosomal recessive 35	602167	AR
EYA1	Branchio-oto-renal syndrome 1	601653	AD
EYA4	Deafness, autosomal dominant 10	603550	AD
FDXR	Auditory neuropathy and optic atrophy	103270	AR
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia (Deafness with LAMM)	164950	AR
FGFR1	Pfeiffer syndrome	136350	AD
FGFR2	Pfeiffer syndrome /Apert syndrome/Crouzon syndrome/LADD syndrome	176943	AD
FGFR3	Camptodactyly, Tall Stature, Scoliosis, and Hearing Loss (CATSHLS) syndrome/LADD syndrome/Muenke syndrome	134934	AD
FITM2	Siddiqi syndrome	612029	AR
FOXI1	Pendred syndrome	601093	AR
GAB1	Deafness, autosomal recessive 26	604439	AR
GATA3	Hypoparathyroidism-deafness-renal disease (HDR syndrome/Barakat syndrome)	131320	AD
GIPC3	Deafness, autosomal recessive 15/Deafness, autosomal recessive 72/Deafness, autosomal recessive 95	608792	AR
GJB2	Deafness, autosomal recessive B1/Deafness, autosomal dominant 3	220290	AD/AR
GJB3	Deafness, autosomal dominant 2B	603324	AD
GJB6 [1]	Deafness, autosomal recessive 1B	604418	AR
GPRASP2	X-linked syndromic hearing loss 7	300969	XLR
GPSM2	Chudley-McCullough syndrome	609245	AR
GRAP	Deafness, autosomal recessive 114	604330	AR
GREB1L	Inner ear malformations and deafness	617782	AD
GRHL2	Deafness, autosomal dominant 28	608576	AD
GRXCR1	Deafness, autosomal recessive 25	613283	AR
GRXCR2	Deafness, autosomal recessive 101	615762	AR
GSDME	Deafness, autosomal dominant 5	608798	AD
HARS2	Perrault syndrome type 2	600783	AR
HGF	Deafness, autosomal recessive 39	142409	AR
HOMER2	Deafness, autosomal dominant 68	604799	AD
HOXA2	Microtia with or without hearing loss	604685	AD/AR
HOXB1	Facial paresis, hereditary congenital, 3	142968	AR
HSD17B4	Perrault syndrome type 1	601860	AR
IFNLR1	Deafness, autosomal dominant 2C	607404	AD
ILDR1	Deafness, autosomal recessive 42	609739	AR
KARS1	Deafness, autosomal recessive 89	601421	AR
KCNE1	Jervell and Lange-Nielsen syndrome 2	176261	AR
KCNJ10	Pendred syndrome	602208	AR
KCNQ1	Jervell and Lange-Nielsen syndrome 1	607542	AR
KCNQ4	Deafness, autosomal dominant 2A	603537	AD
KITLG	Deafness, autosomal dominant 69/Waardenburg syndrome 2	184745	AD
KMT2D	Kabuki syndrome 1	602113	AD
LARS2	Perrault syndrome type 4	604544	AR
LHFPL5	Deafness, autosomal recessive 67	609427	AR
LHX3	Pituitary hormone deficiency, combined	600577	AR
LMX1A	Deafness, autosomal dominant 7/Deafness, autosomal recessive*	600298	AD/AR
LOXHD1	Deafness, autosomal recessive 77	613072	AR
LOXL3	Stickler syndrome	607163	AR
LRP2	Donnai-Barrow syndrome	600073	AR
LRTOMT	Deafness, autosomal recessive 63	612414	AR
MAN2B1	Mannosidosis, Alpha B, lysosomal	609458	AR
MANBA	Mannosidosis, Beta A, lysosomal	609489	AR
MARVELD2	Deafness, autosomal recessive 49	610572	AR
MASP1	3MC syndrome 1	600521	AR
MCM2	Deafness, autosomal dominant 70	116945	AD
MET	Deafness, autosomal recessive 97	164860	AR
MGP	Keutel syndrome	154870	AR
MIR96	Deafness, autosomal dominant 50	611606	AD
MITF	Waardenburg syndrome, type 2A	156845	AD
MPZL2	Deafness, autosomal recessive 111	604873	AR
MSRB3	Deafness, autosomal recessive 74	613719	AR
MT-CO1	Deafness, non-syndromic senorineural, mitochondrial	516030	M
MT-ND1	MELAS/Deafness, nonsyndromic sensorineural, mitochondrial	516000	M
MT-RNR1	Deafness, aminoglycoside-induced	561000	M
MT-TH	MERRF-MELAS overlap syndrome/Deafness, non-syndromic sensorineural, mitochondrial	590040	M

MT-TI	Cardiomyopathy, familial hypertrophic, deafness, nonsyndromic sensorineural, mitochondrial	590045	M
MT-TK	MERRF/MEERF-MELAS overlap/Cardiomyopathy and deafness/MIDD	590060	M
MT-TL1	MELAS/MERRF	590050	M
MT-TS1	MERRF-MELAS overlap syndrome/Keratoderma, palmoplantar, with deafness/Deafness, nonsyndromic sensorineural, mitochondrial	590080	M
MT-TS2	Cerebellar ataxia, cataract, and diabetes mellitus/MEERF-MELAS overlap syndrome	590085	M
MYH14	Deafness, autosomal dominant 4A	608568	AD
MYH9	Deafness, autosomal dominant 17/Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	160775	AD
MYO15A	Deafness, autosomal recessive 3	602666	AR
MYO3A	Deafness, autosomal recessive 30/Deafness, autosomal dominant*	606808	AD/AR
MYO6	Deafness, autosomal dominant 22/Deafness, autosomal recessive 37	600970	AD/AR
MYO7A	Deafness, autosomal dominant 11/Deafness, autosomal recessive 2/Usher syndrome type 1B	276903	AD/AR
NARS2	Deafness, autosomal recessive 94/Combined oxidative phosphorylation deficiency 24	612803	AR
NDP	Norrie disease	300658	XLR
NEFL	Charcot-Marie-Tooth disease type 2E/Charcot-Marie-Tooth disease, demyelinating, type 1F/Charcot-Marie-Tooth disease, dominant intermediate G	162280	AD/AR
NF2	Neufibromatosis, type 2	607379	AD
NLRP3	Muckle-Wells syndrome, Neonatal Onset Multisystem Inflammatory Disease (NOMID); Chronic Infantile Neurologic Cutaneous and Articular (CINCA) syndrome (Cryopyrin-associated periodic syndrome (CAPS))	606416	AD
NOG	Stapes ankylosis with broad thumbs and toes	602991	AD
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome	132890	AD
OPA1	Optic atrophy plus syndrome	605290	AD
OSBPL2	Deafness, autosomal dominant 67	606731	AD
OTOA	Deafness, autosomal recessive 22	607038	AR
OTOF	Deafness, autosomal recessive 9/Auditory neuropathy, autosomal recessive 1	603681	AR
OTOG	Deafness, autosomal recessive 18B	604487	AR
OTOGL	Deafness, autosomal recessive 84B	614925	AR
P2RX2	Deafness, autosomal dominant 41	600844	AD
PAX1	Otofaciocervical syndrome 2	167411	AR
PAX3	Waardenburg syndrome type 1/Waardenburg syndrome type 3	606597	AD/AR
PCDH15	Deafness, autosomal recessive 23/Usher Syndrome type 1F	605514	AR
PDE1C	Deafness, autosomal dominant 74	602987	AD
PDZD7	Deafness, autosomal recessive 57	612971	AR
PEX1	Heimler syndrome 1/Peroxisome biogenesis disorder 1A/Peroxisome biogenesis disorder 1B (Zellweger spectrum disorders)	602136	AR
PEX26	Peroxisome biogenesis disorder 7A/Peroxisome biogenesis disorder 7B (Zellweger spectrum disorders)	608666	AR
PEX6	Heimler syndrome 2/Peroxisome biogenesis disorder 4A/Peroxisome biogenesis disorder 4B (Zellweger spectrum disorders)	601498	AR
PJVK	Deafness, autosomal recessive 59	610219	AR
PLS1	Deafness, autosomal dominant 76	602734	AD
PNPT1	Deafness, autosomal recessive 70	610316	AR
POLR1B	Treacher-Collins syndrome 4	602000	AD
POLR1C	Treacher-Collins syndrome 3	610060	AR
POLR1D	Treacher-Collins syndrome 2	613715	AD
POU3F4	Deafness, X-linked 2	300039	XLR
POU4F3	Deafness, autosomal dominant 15	602460	AD
PPIP5K2	Deafness, autosomal recessive 100	611648	AR
PRPS1	Deafness, X-linked 1/Arts syndrome/Charcot Marie Tooth X-linked recessive, 5/ Phosphoribosylpyrophosphate synthetase superactivity *(PRPS1* -related disorders)	311850	XLR
PTPRQ	Deafness, autosomal dominant 73/Deafness, autosomal recessive 84A	603317	AD/AR
RAI1	Smith-Magenis syndrome	607642	AD
RDX	Deafness, autosomal recessive 24	179410	AR
REST	Deafness, autosomal dominant 27	600571	AD
RIPOR2	Deafness, autosomal dominant 21/Deafness, autosomal recessive 104	611410	AD/AR
ROR1	Deafness, autosomal recessive 108	602336	AR
S1PR2	Deafness, autosomal recessive 68	605111	AR
SEMA3E	CHARGE syndrome	608166	AD
SERPINB6	Deafness, autosomal recessive 91	173321	AR
SIX1	Deafness, autosomal dominant 23/Branchio-oto-renal syndrome 3	601205	AD
SIX2	Ptosis, frontonasal dysplasia, and conductive hearing loss	604994	AD
SIX5	Branchio-Oto-Renal syndrome 2	600963	AD
SLC17A8	Deafness, autosomal dominant 25	607557	AD
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	603941	AR
SLC22A4	Deafness, autosomal recessive 60	604190	AR
SLC26A4	Deafness, autosomal recessive 4/Pendred syndrome	605646	AR
SLC26A5	Deafness, autosomal recessive 61	604943	AR
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration	603690	AR

SLC44A4	Deafness, autosomal dominant 72	606107	AD
SLC4A11	Corneal dystrophy and perceptive deafness (Harboyan syndrome)	610206	AR
SLC52A2	Brown-Vialetto-Van Laere syndrome 2	607882	AR
SLC52A3	Brown-Vialetto-Van Laere syndrome 1	613350	AR
SLITRK6	Deafness and myopia	609681	AR
SMPX	Deafness, X-linked 4	300226	XLD
SNAI2	Waardenburg syndrome type 2D	602150	AD
SOX10	Waardenburg syndrome type 2E/4C	602229	AD
SPATA5	Epilepsy, hearing loss, and mental retardation syndrome	613940	AR
SPNS2	Deafness, autosomal recessive 115	612584	AR
STRC	Deafness, autosomal recessive 16/Deafness infertility syndrome	606440	AR
SUCLA2	Mitochondrial DNA depletion syndrome 5	603921	AR
SYNE4	Deafness, autosomal recessive 76	615535	AR
TBC1D24	Deafness, autosomal dominant 65/Deafness, autosomal recessive 86/DOORS syndrome	613577	AD/AR
TBL1X	Hypothyroidism, congenital, nongoitrous, 8	300196	XLR
TBX1	Velocardiofacial syndrome	602054	AD
TCOF1	Treacher-Collins syndrome	606847	AD
TECTA	Deafness, autosomal dominant 8/Deafness, autosomal dominant 12/Deafness, autosomal recessive 21	602574	AD/AR
TFAP2A	Branchiooculofacial syndrome	107580	AD
TIMM8A	Mohr-Tranebjaerg syndrome (DDON syndrome)	300356	XLR
TJP2	Deafness, autosomal dominant 51	607709	AD
TMC1	Deafness, autosomal dominant 36/Deafness, autosomal recessive 7/Deafness, autosomal recessive 11	606706	AD/AR
TMEM126A	Optic atrophy 7 with or without auditory neuropathy	612988	AR
TMEM132E	Deafness, autosomal recessive 99	616178	AR
TMIE	Deafness, autosomal recessive 6	607237	AR
TMPRSS3	Deafness, autosomal recessive 8/Deafness, autosomal recessive 10	605511	AR
TNC	Deafness, autosomal dominant 56	187380	AD
TPRN	Deafness, autosomal recessive 79	613354	AR
TRIOBP	Deafness, autosomal recessive 28	609761	AR
TRRAP	Deafness, autosomal dominant*	603015	AD
TSPEAR	Deafness, autosomal recessive 98	612920	AR
TUBB4B	Leber congenital amaurosis with early-onset deafness	602660	AD
TWNK	Perrault syndrome type 5	606075	AR
USH1C	Deafness, autosomal recessive 18A/Usher syndrome type 1C	605242	AR
USH1G	Usher syndrome type 1G	607696	AR
USH2A	Usher syndrome type 2A	608400	AR
WBP2	Deafness, autosomal recessive 107	606962	AR
WFS1	Deafness, autosomal dominant 6/Deafness, autosomal dominant 14/Deafness, autosomal dominant 38/Wolfram syndrome 1	606201	AD/AR
WHRN	Deafness, autosomal recessive 31/Usher syndrome type 2D	607928	AR

[1]Genomic Regions covered for copy number variant analysis only. Additional genomic regions covered for copy number analysis: *CATSPER2*[1], *CRYL1*[1], *OTOAP1*[1], *STRCP1*[1]

*Locus number not yet assigned, AD=autosomal dominant, AR=autosomal recessive, XLD=X-linked dominant, XLR=X-linked recessive, M=mitochondrial

Some genes on OtoSCOPE v9 are associated with additional disorders that do not include hearing loss or deafness as a phenotype. These disorders are not included in the list above.

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