Molecular Otolaryngology & Renal Research Laboratories
The University of Iowa • 5270 CBRB • Iowa City, IA 52242-1078
319-335-7997 Tel • 319-353-5869 Fax • https://morl.lab.uiowa.edu/

OtoSCOPE® v8 (152 Gene) List

Autosomal Recessive Genes	
Gene	**Locus**
ADCY1	DFNB44
BDP1	DFNB*
BSND	DFNB73;BARTS4A
CABP2	DFNB93
CDC14A	DFNB105/32;HIIMS
CDH23	DFNB12;USH1D
CIB2	DFNB48
CLDN14	DFNB29
CLIC5	DFNB103
COL11A2	DFNB53;DFNA13;STL3
DCDC2	DFNB66
ELMOD3	DFNB88
EPS8	DFNB102
EPS8L2	DFNB106
ESPN	DFNB36
ESRRB	DFNB35
FAM65B	DFNB104
GIPC3	DFNB15/72/95
GJB2	DFNB1;DFNA3
GJB6	DFNB1B;DFNA3B
GRXCR1	DFNB25
GRXCR2	DFNB101
HGF	DFNB39
ILDR1	DFNB42
KARS	DFNB89
LHFPL5	DFNB67
LOXHD1	DFNB77
LRTOMT	DFNB63
MARVELD2	DFNB49
MET	DFNB97
MSRB3	DFNB74
MYO3A	DFNB30;DFNA*
MYO6	DFNB37;DFNA22
MYO7A	DFNB2;DFNA11;USH1B
MYO15A	DFNB3
NARS2	DFNB94
OTOA	DFNB22
OTOF	DFNB9
OTOG	DFNB18B
OTOGL	DFNB84B
PCDH15	DFNB23;USH1F
PDZD7	DFNB57
PJVK	DFNB59
PNPT1	DFNB70
PTPRQ	DFNB84A;DFNA73
RDX	DFNB24
ROR1	DFNB108
S1PR2	DFNB68
SERPINB6	DFNB91
SLC22A4	DFNB60
SLC26A4	DFNB4;PDS
SLC26A5	DFNB61
STRC	DFNB16
SYNE4	DFNB76
TBC1D24	DFNB86;DFNA65
TECTA	DFNB21;DFNA8/12
TMC1	DFNB7/11;DFNA36
TMEM132E	DFNB99
TMIE	DFNB6
TMPRSS3	DFNB8/10
TPRN	DFNB79
TRIOBP	DFNB28
TSPEAR	DFNB98
USH1C	DFNB18A;USH1C
WHRN	DFNB31;USH2D
Total	**65**

Autosomal Dominant Genes	
Gene	**Locus**
ACTG1	DFNA20/26
CCDC50	DFNA44
CD164	DFNA66
CEACAM16	DFNA4B
COCH	DFNA9
COL11A1	DFNA37;STL2
COL11A2	DFNA13;DFNB53;STL3
CRYM	DFNA40
DFNA5	DFNA5
DIABLO	DFNA64
DIAPH1	DFNA1
EYA4	DFNA10
GJB2	DFNA3;DFNB1
GJB3	DFNA2B
GJB6	DFNA3B;DFNB1B
GRHL2	DFNA28
HOMER2	DFNA68
KCNQ4	DFNA2A
KITLG	DFNA69;WS2
MCM2	DFNA70
MiR-96	DFNA50
MYH14	DFNA4A
MYH9	DFNA17
MYO3A	DFNA*/DFNB30
MYO6	DFNA22;DFNB37
MYO7A	DFNA11;DFNB2;USH1B
OSBPL2	DFNA67
P2RX2	DFNA41
PTPRQ	DFNA73;DFNB84A
POU4F3	DFNA15
SIX1	DFNA23;BOR3
SLC17A8	DFNA25
TBC1D24	DFNA65;DFNB86
TECTA	DFNA8/12;DFNB21
TECTB	DFNA*
TJP2	DFNA51
TMC1	DFNA36;DFNB7/11
TNC	DFNA56
WFS1	DFNA6/14/38;WFS1/WFSL
Total	**39**

X-linked	
Gene	**Locus**
COL4A6	DFNX6 (XLR)
POU3F4	DFNX2 (XLR)
PRPS1	DFNX1 (XL)
SMPX	DFNX4 (XLD)
Total	**4**

OtoSCOPE® Version 8 (152 gene)	
Size	1.208 Mbp

Several genes exhibit pleiotropy and are causally linked to both AD and AR or syndromic and non-syndromic hearing loss. Genes on the OtoSCOPE® v8 platform are evaluated for single nucleotide and copy number variants.

Alport Syndrome	
COL4A3	ATS (AR/AD)
COL4A4	ATS (AR)
COL4A5	ATS (XR)
Total	**3**

Alström Syndrome	
ALMS1	ALMS1 (AR)
Total	**1**

Audiogenic Seizures	
GIPC3	(AR)
Total	**1**

Auditory Neuropathy	
AIFM1	AUNX1 (XR)
DIAPH3	AUNA1 (AD)
OTOF	DFNB9 (AR)
PJVK	DFNB59 (AR)
Total	**4**

Branchio-Oto-Renal Syndrome	
EYA1	BOR1 (AD)
SIX1	BOR3 (AD)
SIX5	BOR2 (AD)
Total	**3**

Chudly-McCullough Syndrome	
GPSM2	CMCS (AR)
Total	**1**

Deafness-Male Infertility	
CDC14A	HIMMS (AR)
STRC-CATSPER2	DIS (AR)
Total	**2**

Deafness & Microdontia	
FGF3	HL & LAMM (AR)
Total	**1**

Dentinogenesis Imperfecta & HL	
DSPP	DGI1 (AD)
Total	**1**

DRTA & HL	
ATP6V1B1	HL & DRTA (AR)
Total	**1**

Heimler Syndrome	
PEX1	HMLR1 (AR)
PEX6	HMLR2 (AR)
Total	**2**

High Myopia & HL	
SLITRK6	DFNMYP (AR)
Total	**1**

Hypoparathyroidism Renal Disease & HL	
GATA3	HDR (AD)
Total	**1**

Jervell & Lange Nielsen	
KCNE1	JLNS2 (AR)
KCNQ1	JLNS1 (AR)
Total	**2**

Mitochondrial	
MTRNR1	
MTTS1	
MTTL1	MELAS
Total	**3**

Modifier Genes	
ATP2B2	DFNB12 modifier
Total	**1**

Mohr-Tranebjaerg Syndrome	
TIMM8A	MTS (XR)
Total	**1**

Muckle-Wells Syndrome	
NLRP3	MWS (AD)
Total	**1**

Optic Atrophy Plus Syndrome	
OPA1	OPA1 (AD)
Total	**1**

OSMED	
COL11A2	OSMEDA/OSMEDB (AR, AD)
Total	**1**

Pendred Syndrome	
FOXI1	PDS (AR)
KCNJ10	PDS (AR)
SLC26A4	PDS (AR)
Total	**3**

Perrault Syndrome	
CLPP	PRLTS3 (AR)
HARS2	PRLTS2 (AR)
HSD17B4	PRLTS1 (AR)
LARS2	PRLTS4 (AR)
TWNK	PRLTS5 (AR)
Total	**5**

Pfeiffer Syndrome	
FGFR1	PS1 (AD)
FGFR2	PS1/PS2/PS3 (AD)
Total	**2**

Sinoatrial Node Dysfunction & HL	
CACNA1D	SANDD (AR)
Total	**1**

Stickler Syndrome	
COL2A1	STL1 (AD)
COL9A1	STL4 (AR)
COL9A2	STL5 (AR)
COL11A1	STL2 (AD)
COL11A2	STL3 (AR/AD)
LOXL3	STL (AR)
Total	**6**

Treacher-Collins Syndrome	
POLR1C	TCS3 (AR)
POLR1D	TCS2 (AD)
TCOF1	TCS1 (AD)
Total	**3**

Usher Syndrome	
ADGRV1	USH2C (AR)
CDH23	USH1D (AR)
CLRN1	USH3A (AR)
MYO7A	USH1B (AR)
PCDH15	USH1F (AR)
USH1C	USH1C (AR)
USH1G	USH1G (AR)
USH2A	USH2A (AR)
WHRN	USH2D (AR)
Total	**9**

Velocardiofacial Syndrome	
TBX1	VCFS (AD)
Totals	**1**

Waardenburg Syndrome	
EDN3	WS4B (AR/AD)
EDNRB	WS4A (AR/AD)
KITLG	WS2 (AD)
MITF	WS2A (AD)
PAX3	WS1/WS3 (AD)
SNAI2	WS2D (AD)
SOX10	WS2E/WS4C (AD)
Total	**7**

Wolfram Syndrome	
CISD2	WFS2 (AR)
WFS1	WFS1 (AR/AD)
Total	**2**

Genomic Regions Covered for CNV Analysis	
CATSPER2	DIS
CRYL1	DFNB1 deletions
LOC653786	*OTOA* pseudogene
STRC promoter	*STRC*
STRCP1	*STRC* pseudogene
Total	**5**

AD=autosomal dominant; **AR**=autosomal recessive; **CNV**=copy number variation; **DIS**=Deafness infertility syndrome, **DRTA**=Distal renal tubular acidosis; **HL**= hearing loss; **LAMM**=Labyrinthine aplasia, microtia, & microdontia; **MELAS**=Mitochondrial encephalomyopathy, lactic acidosis, & stroke-like episodes; **OSMED=**Otospondylomegaepiphyseal dysplasia; **XR**=X-linked recessive.*

This test was developed and its performance characteristics determined by the Clinical Diagnostics Division of the Molecular Otolaryngology & Renal Research Laboratories. It has not been cleared or approved by the US Food & Drug Administration. (21 CFR§ 809.30 [e])