OtoGenome Gene List							
Gene	Evid.	Inher.	Mutation Spect.	NonSynd.	Synd.	Phenotype	
						General Hearing Loss Phenotype	Associated Syndrome/Other Features
ACTG1	3	AD	M	X[3]	X[2]	Postlingual progressive SNHL	Baraitser-Winter syndrome
ADCY1	2	AR	LOF	X		Mild to severe mixed hearing loss	
ADGRV1	3	AR	M, LOF		X[3]	Prelingual moderate to profound sloping SNHL	Usher syndrome type 2
ALMS1	3	AR	M, LOF		X	Progressive SNHL	Alstrom syndrome
ATP6V1B1	3	AR	M, LOF		X[3]	Childhood onset progressive SNHL	Distal renal tubular acidosis
BCS1L	3	AR	M, LOF		X[3]	Congenital SNHL with variable severity	Bjornstad syndrome
BSND	3	AR	M, LOF	X[2]	X[3]	Prelingual severe to profound SNHL	Bartter syndrome
CABP2	3	AR	M, LOF	X		Prelingual moderate to severe SNHL	
CACNA1D	2	AR	LOF		X[2]	Congenital severe to profound flat SNHL	Bradycardia and deafness
CATSPER2[$]	3	AR			X[3]	N/A	Deafness male infertility syndrome (DIS)
CCDC50	2	AD	LOF	X[2]		Postlingual progressive moderate to profound SNHL	
CDC14A	2	AR	LOF	X		Congenital severe to profound SNHL	
CDH23	3	AR	M, LOF	X[3]	X[3]	Congenital moderate to profound SNHL	Usher syndrome type 1
CD164	2	AD	LOF	X		Moderate to severe SNHL with variable onset (congenital to early 20s)	
CEACAM16	2	AD	M	X[2]		Postlingual progressive moderate SNHL	
CEP78	3	AR	M, LOF		X	SNHL with postlingual onset	Cone-rod dystrophy
CHD7	3	AD	M, LOF		X	Mixed or sensorineural hearing loss often with temporal bone abnormalities	CHARGE syndrome
CIB2	3	AR	M	X[3]	X[1]	Prelingual severe to profound flat SNHL	Usher syndrome type 1
CLDN14	3	AR	M, LOF	X[3]		Prelingual flat SNHL (variable progression)	
CLIC5	2	AR	LOF	X		Progressive SNHL	Vestibular dysfunction
CLPP	3	AR	M, LOF		X[3]	Congenital severe to profound flat SNHL	Perrault syndrome
CLRN1	3	AR	M, LOF		X[3]	Variable onset progressive moderate to severe SNHL	Usher syndrome type 3
COCH	3	AD	M	X[3]		Postlingual progressive profound SNHL	Vestibular impairment
COL4A3	3	AR, AD	M, LOF		X	Progressive SNHL with onset typically in late childhood early adolescence	Alport syndrome
COL4A4	3	AR, AD	M, LOF		X	Progressive SNHL with onset typically in late childhood early adolescence	Alport syndrome
COL4A5	3	XLD	M, LOF		X	Progressive sensorineural hearing loss with onset typically in late childhood, early adolescence	Alport syndrome
COL11A2	3	AD[3]	M, In-frame del	X[3]		Congenital mild to moderately severe mid-frequency SNHL	
					X[3]	Childhood/adulthood onset mild to moderate SNHL	Non-ocular Stickler syndrome (STL3)
		AR[3]	M, LOF	X[1-2]		Prelingual profound flat/mid frequency SNHL	
					X[3]	Childhood moderate to profound flat SNHL	OSMED syndrome
DIABLO	3	AD	M	X[3]		Adulthood onset progressive mild to moderate flat SNHL	
DFNA5	2	AD	Exon 8 skipping	X[2]		Postlingual progressive sloping SNHL	
DFNB59	3	AR	M, LOF	X[3]		Prelingual severe to profound flat SNHL	Auditory neuropathy
DIAPH1	3	AD	M, LOF	X[3]		Postlingual low frequency progressive SNHL	
EDN3	3	AD/AR	M, LOF		X[3]	Variable	Waardenburg syndrome type 4
EDNRB	3	AD/AR	M, LOF		X[3]	Variable	Waardenburg syndrome type 4
EPS8	2	AR	LOF	X		Congenital profound SNHL	
ESPN	3	AD[1], AR[3]	LOF	X[3]		Prelingual severe to profound flat SNHL	Vestibular areflexia, in some
ESRRB	3	AR	M*, LOF	X[3]		Early onset severe to profound flat/slightly sloping SNHL	
EYA1	3	AD	M, LOF		X[3]	Variable onset mild to profound SNHL	Branchio-oto-renal syndrome
EYA4	3	AD	LOF	X[3]		Postlingual progressive moderate to profound flat SNHL	
GIPC3	3	AR	M, LOF	X[3]		Prelingual mild to profound flat SNHL	
GJB2	3	AD[3]	M	X[2-3]		Congenital/late onset mild to profound SNHL	
					X[2-3]	Childhood onset moderate to severe sloping SNHL	Dermatologic manifestations

		AR[3]	M, LOF	X[3]		Congenital/childhood onset mild to profound SNHL	
GJB6	3	AR	D13S1830 del	X[3]		Congenital/childhood onset mild to profound SNHL	GJB2 downregulation
		AD	M		X[1]	_	Hidrotic Ectodermal Dysplasia
			M, LOF	X[1]		Variable	
GPSM2	3	AR	LOF		X[3]	Prelingual severe to profound slightly sloping SNHL	Chudley-McCullough syndrome
GRHL2	3	AD	LOF	X[3]		Postlingual progressive mild to severe SNHL	
GRXCR1	2	AR	M, LOF	X[2]		Congenital moderate to profound flat/slightly sloping SNHL	
HARS	1-2	AR	M		X[1-2]	Childhood onset progressive SNHL	Usher syndrome type 3B
HARS2	2	AR	M		X[2]	Variable onset progressive mild to severe flat SNHL	Perrault syndrome
HGF	2	AR	Intronic del, splice	X[2]		Prelingual severe to profound sloping SNHL	
HSD17B4	2	AR	M, LOF		X[2]	Childhood onset moderate to severe SNHL	Perrault syndrome
ILDR1	3	AR	M, LOF*	X[3]		Prelingual moderate to profound sloping SNHL	
KARS	2	AR	M	X[2]	X[2]	Prelingual moderate to severe flat SNHL	Peripheral neuropathy
KCNE1	3	AR	M		X[3]	Congenital severe to profound flat SNHL	Jervell and Lange-Nielsen syndrome
KCNQ1	3	AR	M, LOF		X[3]	Congenital severe to profound flat SNHL	Jervell and Lange-Nielsen syndrome
KCNQ4	3	AD	M, LOF	X[3]		Postlingual progressive sloping SNHL	
KITLG	2	AD	M, LOF	X	X	Variable SNHL	Waardenburg syndrome type 2, hyperpigmentation
LARS2	2	AR	M, LOF		X[2]	Childhood onset progressive mild to severe low frequency SNHL	Perrault syndrome
LHFPL5	3	AR	M, LOF	X[3]		Prelingual severe to profound SNHL	
LOXHD1	3	AR	M, LOF*	X[3]	X[1]	Variable onset variable SNHL	Fuchs corneal dystrophy
LRTOMT	3	AR	M, LOF	X[3]		Congenital moderate to profound flat SNHL	
MARVELD2	3	AR	LOF	X[3]		Prelingual moderate to profound flat/sloping SNHL	
MIR96	3	AD	Seed region	X[3]	X[1]	Postlingual progressive flat/sloping SNHL	Vertigo in some
MITF	3	AD	M, LOF		X[3]	Variable	Waardenburg syndrome type 2
MSRB3	2	AR	M, LOF	X[2]		Prelingual severe to profound flat SNHL	
MTRNR1	3	Mito.	Point mutat.	X[3]		Variable progressive SNHL	Aminoglycoside ototoxicity sensitivity
MTTS1	3	Mito.	Point mutat.	X[3]		Variable progressive SNHL	
MYH14	3	AD	M*, LOF	X[3]	X[1]	Postlingual moderate to profound flat SNHL	Peripheral neuropathy
MYH9	3	AD	M*, LOF	X[2]	X[3]	Variable onset progressive SNHL	Macrothrombocytopenia, MYH9-related disorder
MYO15A	3	AR	M, LOF	X[3]		Congenital severe to profound flat SNHL	
MYO3A	3	AR	LOF	X[3]		Postlingual progressive moderate to severe sloping SNHL	
MYO6	3	AD[3]	M, LOF	X[3]		Postlingual progressive moderate to profound sloping SNHL	
		AR[3]	LOF	X[3]		Congenital profound SNHL	Vestibular impairment in some
MYO7A	3	AR	M, LOF		X[3]	Congenital severe to profound flat SNHL	Usher syndrome type 1
			M, LOF	X[2]		Congenital severe to profound flat SNHL	Vestibular impairment
		AD	M, In-frame del	X[2]		Postlingual mild to severe SNHL	Vestibular impairment
NLRP3	3	AD	M		X	Conductive mixed or sensorineural hearing loss	Cryopyrin-associated periodic syndrome (CAPS)
OSBPL2	2	AD	M, del	X		Postlingual progressive SNHL	
OTOA	3	AR	M, LOF	X[3]		Prelingual severe to profound flat SNHL	
OTOF	3	AR	M, LOF	X[3]		Congenital severe to profound flat SNHL	Auditory neuropathy
OTOG	2	AR	M, LOF	X[2]		Prelingual/childhood onset moderate flat/slightly sloping SNHL	Vestibular impairment in some
OTOGL	3	AR	LOF	X[3]		Congenital moderate to moderately severe sloping SNHL	
P2RX2	3	AD	M	X[3]		Adolescent onset progressive moderately severe flat SNHL	High frequency tinnitus
PAX3	3	AD	M, LOF		X[3]	Variable	Waardenburg syndrome types 1 and 3
PCDH15	3	AR	M, LOF	X[3]	X[3]	Congenital profound flat SNHL	Usher syndrome type 1

PDZD7	3	AR		X		Prelingual mild to profound SNHL	
POU3F4	3	X-linked	M, LOF	X[3]		Congenital moderate to profound flat mixed HL	IAC dilation, perilymphatic gusher
POU4F3	3	AD	M, LOF	X[3]		Adult onset progressive moderate to severe sloping SNHL	
PRPS1	3	X-linked	M	X[3]	X[3]	Postlingual progressive severe to profound flat SNHL	PRS-I/Arts/CMT
RDX	3	AR	M, LOF	X[3]		Prelingual severe to profound flat SNHL	
RIPOR2	3	AR	LOF	X		Prelingual profound SNHL	
S1PR2	3	AR	M	X		Congenital profound SNHL	
SERPINB6	2	AR	LOF	X[2]		Postlingual moderate to severe sloping SNHL	
SIX1	3	AD	M, LOF		X[3]	Variable (3wk-22y) onset mild to severe mixed HL	Branchio-oto-renal syndrome
SLC26A4	3	AR	M, LOF	X[3]	X[3]	Congenital progressive severe to profound SNHL with temporal bone abnormalities	Pendred syndrome
SLC52A2	3	AR	M		X	Variable onset and severity of SNHL	Brown-Vialetto-Van Laere syndrome
SLC52A3	2	AR	M, LOF		X	Progressive SNHL with onset from infancy to adolescence	Brown-Vialetto-Van Laere syndrome
SLITRK6	3	AR	LOF		X	Congenital moderate to severe SNHL	High myopia
SMPX	3	X-linked	LOF	X[3]		Postlingual progressive moderate to profound flat/sloping SNHL	
SNAI2	1-2	AR	del		X	Severe/profound HL	Waardenburg syndrome type 2
SOX10	3	AD	M, LOF		X[3]	Variable HL	Waardenburg syndrome types 2 and 4
STRC	3	AR	M, LOF, del	X[3]	X[3]	Childhood onset mild to moderate sloping or mid frequency SNHL	Deafness male infertility syndrome (DIS)
SYNE4	2	AR	LOF	X[2]		Pre/postlingual progressive mild to profound sloping SNHL	
TBC1D24	3	AR	M	X[3]	X[3]	Prelingual profound flat SNHL	Epilepsy/DOOR syndrome
	2	AD	M	X[2]		Progressive sloping moderate to severe SNHL	
TECTA	3	AD[3]	M	X[3]		Pre/postlingual progressive (in some) mild to severe SNHL	
		AR[3]	LOF	X[3]		Prelingual moderate to profound high/mid frequency SNHL	
TIMM8A	3	X-linked	M, LOF*		X	Congenital/early childhood onset progressive profound flat SNHL	Mohr-Tranebjaerg syndrome
TMC1	3	AD[3]	M	X[3]		Postlingual progressive SNHL	
		AR[3]	LOF	X[3]		Congenital profound flat/slightly sloping SNHL	
TMIE	3	AR	M, LOF	X[3]		Congenital severe to profound flat SNHL	
TMPRSS3	3	AR	M, LOF	X[3]		Congenital/childhood onset severe to profound flat SNHL	
TPRN	3	AR	LOF	X[3]		Prelingual severe to profound flat/slightly sloping SNHL	
TRIOBP	3	AR	LOF	X[3]		Prelingual severe to profound flat SNHL	
USH1C	3	AR	M, LOF	X[3]	X[3]	Prelingual severe to profound flat SNHL	Usher syndrome type 1
USH1G	3	AR	M, LOF*		X[3]	Congenital profound flat SNHL	Usher syndrome type 1
USH2A	3	AR	M, LOF		X[3]	Prelingual moderate to profound sloping SNHL	Usher syndrome type 2
		AR	M, LOF	X[3]		N/A	Isolated autosomal recessive retinitis pigmentosa
WFS1	3	AD[3]	M	X[3]		Congenital slowly progressive low frequency SNHL	
					X[2]	Childhood onset progressive mild to moderate low to mid frequency SNHL	Wolfram-like disorder
		AR[3]	M, LOF		X[3]	Early onset progressive sloping SNHL	Wolfram syndrome
WHRN	3	AR	M, LOF*	X[3]	X[3]	Prelingual moderate to profound sloping SNHL	Usher syndrome type 2

Key:

1 - Weak Association

2 - Likely Association

3 - Definitive Association

* - Most common

$ - Gene is not sequenced - included for deletion/duplication purposes only