HEARING IMPAIRMENT GENE PANEL

Gene symbol	*Depth (reads)*	*Coverage (avg %)*	*OMIM disease*	*Description*
ACTB	101	17	243310	Baraitser-Winter syndrome 1
ACTG1	113	16	614583	Baraitser-Winter syndrome 2
ATP6V1B1	113	97	200	-
BSND	109	89	602522	Bartter syndrome type 4a
CABP2	74	74	614899	Deafness autosomal recessive 93
CACNA1D	126	87	614896	Sinoatrial node dysfunction and deafness
CC2D2A	86	85	216360	COACH syndrome
CCDC50	136	90	607453	Deafness autosomal dominant 44
CDH23	92	88	601386	Deafness autosomal recessive 12
CEACAM16	91	86	614614	Deafness autosomal dominant 4B
CIB2	76	79	609439	Deafness autosomal recessive 48
CLDN14	61	100	614035	Deafness autosomal recessive 29
CLRN1	172	98	614180	Retinitis pigmentosa 61
COCH	139	93	601369	Deafness autosomal dominant 9
COL11A1	98	96	228520	Fibrochondrogenesis
COL11A2	15	85	601868	Deafness autosomal dominant 13
COL4A3	82	83	104200	Alport syndrome autosomal dominant
COL4A4	100	83	203780	Alport syndrome autosomal recessive
COL4A5	64	87	301050	Alport syndrome
COL9A1	110	91	614135	Epiphyseal dysplasia multiple 6
CRYM	72	87	200	Deafness
DFNA5	94	85	600994	Deafness autosomal dominant 5
DFNB31	79	87	611383	Usher syndrome, type 2D
DIAPH1	95	92	124900	Deafness autosomal dominant 1
DIAPH3	105	95	609129	Auditory neuropathy autosomal dominant 1
DSPP	227	26	605594	Deafness autosomal dominant 36 with dentinogenesis
EDN3	93	83	209880	Central hypoventilation syndrome congenital
EDNRB	148	95	600501	ABCD syndrome

ESPN	47	48	609006	Deafness autosomal recessive 36
ESRRB	64	88	608565	Deafness autosomal recessive 35
EYA1	125	82	113650	Anterior segment anomalies with or without cataract
EYA4	126	93	605362	Cardiomyopathy dilated 1J
FGF3	69	100	610706	Deafness congenital with inner ear agenesis microtia and microdontia
FOXI1	91	95	600791	Enlarged vestibular aqueduct
GIPC3	93	90	601869	Deafness autosomal recessive 15
GJB2	161	100	149200	Bart-Pumphrey syndrome
GJB3	135	100	612644	Deafness autosomal dominant 2B
GJB6	177	100	612643	Deafness autosomal dominant 3B
GPR98	120	93	604352	Febrile seizures familial 4
GPSM2	144	92	604213	Chudley-McCullough syndrome
GRHL2	111	90	608641	Deafness autosomal dominant 28
GRXCR1	194	93	613285	Deafness autosomal recessive 25
HARS	139	89	614504	Usher syndrome type 3B
HARS2	168	85	614926	Perrault syndrome 2
HGF	106	97	608265	Deafness autosomal recessive 39
HSD17B4	103	89	261515	D-bifunctional protein deficiency
ILDR1	62	92	609646	Deafness autosomal recessive 42
KCNE1	177	100	612347	Jervell and Lange-Nielsen syndrome 2
KCNJ10	158	94	600791	Enlarged vestibular aqueduct digenic
KCNQ1	64	92	607554	Atrial fibrillation familial 3
KCNQ4	93	91	600101	Deafness autosomal dominant 2A
LHFPL5	168	96	610265	Deafness autosomal recessive 67
LOXHD1	116	88	613079	Deafness autosomal recessive 77
LRTOMT	84	90	611451	Deafness autosomal recessive 63
MARVELD2	157	91	610153	Deafness autosomal recessive 49
MIR96	70	100	613074	Deafness autosomal dominant 50
MITF	135	87	103500	Tietz albinism-deafness syndrome
MSRB3	117	75	613718	Deafness autosomal recessive 74
MYH14	75	79	600652	Deafness autosomal dominant 4A
MYH9	100	85	603622	Deafness autosomal dominant 17
MYO15A	95	92	600316	Deafness autosomal recessive 3

MYO1A	123	84	607841	Deafness autosomal dominant 48
MYO3A	108	92	607101	Deafness autosomal recessive 30
MYO6	105	94	606346	Deafness autosomal dominant 22
MYO7A	82	87	601317	Deafness autosomal dominant 11
NLRP3	128	96	607115	CINCA syndrome
OPA1	121	99	165500	Optic atrophy 1
OTOA	119	83	607039	Deafness autosomal recessive 22
OTOF	95	80	601071	Auditory neuropathy autosomal recessive 1
OTOGL	115	90	614944	Deafness autosomal recessive 84B
PAX3	109	89	122880	Craniofacial-deafness-hand syndrome
PCDH15	129	94	609533	Deafness autosomal recessive 23
PDZD7	85	86	605472	Usher syndrome type IIC GPR98/PDZD7 digenic
PNPT1	96	84	614932	Combined oxidative phosphorylation deficiency 13
POU3F4	123	100	304400	Deafness X-linked 2
POU4F3	153	100	602459	Deafness autosomal dominant 15
PRPS1	116	73	301835	Arts syndrome
PTPRQ	118	94	613391	Deafness autosomal recessive 84A
RDX	61	56	611022	Deafness autosomal recessive 24
SERPINB6	130	98	613453	Deafness autosomal recessive 91
SIX1	80	94	608389	Brachiootic syndrome 3
SIX5	52	93	610896	Branchiootorenal syndrome 2
SLC17A8	131	90	605583	Deafness autosomal dominant 25
SLC26A4	108	89	600791	Deafness autosomal recessive 4 with enlarged vestibular aqueduct
SLC26A5	119	80	613865	Deafness autosomal recessive 61
SMPX	100	82	300066	Deafness X-linked 4
SNAI2	76	100	172800	Piebaldism
SOX10	55	89	609136	PCWH syndrome
STRC	132	38	603720	Deafness autosomal recessive 16
TECTA	125	90	601543	Deafness autosomal dominant 8/12
TIMM8A	56	60	200	Deafness X-linked 1
TJP2	94	84	607748	Hypercholanemia familial
TMC1	117	91	606705	Deafness autosomal dominant 36
TMIE	65	91	600971	Deafness autosomal recessive 6

TMPRSS3	91	87	601072	Deafness autosomal recessive 8/10
TPRN	35	100	613307	Deafness autosomal recessive 79
TRIOBP	125	52	609823	Deafness autosomal recessive 28
TSPEAR	118	94	614861	Deafness autosomal recessive 98
USH1C	89	90	602092	Deafness autosomal recessive 18A
USH1G	103	93	606943	Usher syndrome, type 1G
USH2A	121	91	613809	Retinitis pigmentosa 39
WFS1	141	92	600965	Deafness autosomal dominant 6/14/38

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association

Ad 2. OMIM phenotype descriptions between {} signify risk factors