

Supplementary Information for:

Genomic Analysis of Inherited Hearing Loss in the Palestinian Population

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Table S1. Genes on the sequencing panel

| Gene (OMIM) | SureDesign input | Inherit | Locus (OMIM) | Syndrome and notes |
|--------------------|-------------------------|----------------|---------------------|---|
| <i>ACTG1</i> | <i>ACTG1</i> | AD | DFNA20/26 | |
| <i>ADCY1</i> | <i>NM_021116</i> | AR | DFNB44 | |
| <i>ADGRV1</i> | <i>ADGRV1</i> | AR | | Usher syndrome, type 2C |
| <i>AIFM1</i> | <i>AIFM1</i> | XR | DFNX5 | Auditory neuropathy |
| <i>ALMS1</i> | <i>ALMS1</i> | AR | | Alstrom syndrome |
| <i>ATP2B2</i> | <i>ATP2B2</i> | AR | | DFNB12 modifier |
| <i>ATP6B1</i> | <i>ATP6V1B1</i> | AR | | Renal tubular acidosis with deafness |
| <i>ATP6V1B2</i> | <i>ATP6V1B2</i> | AD | | Deafness with onychodystrophy |
| <i>BCS1L</i> | <i>BCS1L</i> | AR | | Bjornstad syndrome |
| <i>BDP1</i> | <i>BDP1</i> | AR | DFNB49 | |
| <i>BSND</i> | <i>BSND</i> | AR | DFNB73 | |
| <i>CABP2</i> | <i>CABP2</i> | AR | DFNB93 | |
| <i>CACNA1D</i> | <i>CACNA1D</i> | AR | | Sinoatrial node dysfunction and deafness |
| <i>CATSPER2</i> | <i>CATSPER2</i> | AR | | Deafness-infertility syndrome |
| <i>CCDC50</i> | <i>CCDC50</i> | AD | DFNA44 | |
| <i>CD151</i> | <i>CD151</i> | AR | | Nephropathy, pretibial epidermolysis bullosa, deafness |
| <i>CD164</i> | <i>CD164</i> | AD | DFNA66 | |
| <i>CDC14A</i> | <i>CDC14A</i> | AR | DFNB105 | Deafness-infertility syndrome |
| <i>CDH23</i> | <i>CDH23</i> | AR | | Usher syndrome, type 1D |
| <i>CEACAM16</i> | <i>CEACAM16</i> | AD | | |
| <i>CHD7</i> | <i>NM_017780</i> | AD | | CHARGE syndrome |
| <i>CIB2</i> | <i>CIB2</i> | AR | DFNB48 | |
| <i>CISD2</i> | <i>CISD2</i> | AR | | Wolfram syndrome |
| <i>CLDN14</i> | <i>CLDN14</i> | AR | DFNB29 | |
| <i>CLIC5</i> | <i>CLIC5</i> | AR | DFNB103 | |
| <i>CLPP</i> | <i>CLPP</i> | AR | DFNB81 | |
| <i>CLRN1</i> | <i>CLRN1</i> | AR | | Perrault syndrome 3 |
| <i>COCH</i> | <i>COCH</i> | AD | DFNA9 | Usher syndrome, type 3A |
| <i>COL11A1</i> | <i>COL11A1</i> | AD | | Stickler syndrome, type II |
| <i>COL11A2</i> | <i>COL11A2</i> | AD,AR | DFNB53 | Otospondylomegaphyseal dysplasia, Stickler syndrome |
| <i>COL2A1</i> | <i>COL2A1</i> | AD | | Stickler syndrome, type I |
| <i>COL4A3</i> | <i>NM_000091</i> | AD,AR | | Alport syndrome |
| <i>COL4A4</i> | <i>COL4A4</i> | AR | | Alport syndrome |
| <i>COL4A5</i> | <i>COL4A5</i> | XR | | Alport syndrome |
| <i>COL4A6</i> | <i>COL4A6</i> | XR | DFNX6 | |
| <i>COL9A1</i> | <i>COL9A1</i> | AD | | Stickler syndrome, type IV |
| <i>COL9A2</i> | <i>COL9A2</i> | AR | | Stickler syndrome, type V |
| <i>CRYL1</i> | <i>CRYL1</i> | AR | | large connexin deletion |
| <i>CRYM</i> | <i>CRYM</i> | AD | | |
| <i>DCDC2</i> | <i>DCDC2</i> | AR | DFNB66 | |
| <i>DFNA5</i> | <i>DFNA5</i> | AD | DFNA5 | |
| <i>DIAPH1</i> | <i>DIAPH1</i> | AD | DFNA1 | |
| <i>DIAPH3</i> | <i>DIAPH3</i> | AD | | Auditory Neuropathy |
| <i>DMXL2</i> | <i>DMXL2</i> | AD | | |
| <i>DNMT1</i> | <i>DNMT1</i> | AD | | Cerebellar ataxia, deafness, and narcolepsy |
| <i>DSPP</i> | <i>DSPP</i> | AD | DFNA39 | Deafness with dentinogenesis |
| <i>EDN3</i> | <i>EDN3</i> | AD,AR | | Waardenburg syndrome, type 4B |
| <i>EDNRB</i> | <i>EDNRB</i> | AD,AR | | Waardenburg syndrome, type 4A |
| <i>ELMOD3</i> | <i>ELMOD3</i> | AR | DFNB88 | |
| <i>EPS8</i> | <i>EPS8</i> | AR | DFNB102 | |
| <i>EPS8L2</i> | <i>EPS8L2</i> | AR | | |
| <i>ERAL1</i> | <i>ERAL1</i> | AR | | Perrault syndrome 6 |
| <i>ERCC2</i> | <i>ERCC2</i> | AR | | Cerebrooculofacioskeletal syndrome 2 |
| <i>ESPN</i> | <i>ESPN</i> | AR | DFNB36 | |
| <i>ESRP1</i> | <i>ESRP1</i> | AR | | |
| <i>ESRRB</i> | <i>ESRRB</i> | AR | DFNB35 | |
| <i>EYA1</i> | <i>EYA1</i> | AD | | Branchio-oto-renal syndrome |
| <i>EYA4</i> | <i>EYA4</i> | AD | DFNA10 | |
| <i>FAM65B</i> | <i>FAM65B</i> | AR | | |
| <i>FGF3</i> | <i>FGF3</i> | AR | | Deafness with inner ear agenesis, microtia, and microdontia |
| <i>FGFR1</i> | <i>FGFR1</i> | AD | | Pfieffer syndrome |
| <i>FGFR2</i> | <i>FGFR2</i> | AD | | Pfieffer syndrome/ Crouzon syndrome |
| <i>FOXII</i> | <i>FOXII</i> | AR | | Pendred syndrome |
| <i>GATA3</i> | <i>GATA3</i> | AD | | Hypoparathyroidism, deafness, renal (HDR) syndrome |
| <i>GIPC3</i> | <i>NM_133261</i> | AR | DFNB15/72/95 | Audiogenic Seizures |
| <i>GJB2</i> | <i>GJB2</i> | AR | DFNB1 | |

| Gene (OMIM) | SureDesign input | Inherit | Locus (OMIM) | Syndrome and notes |
|--------------------|------------------------------|----------------|---------------------|--|
| <i>GJB2 exon 1</i> | chr13:20,766,864-20,767,146 | AR | | |
| <i>GJB3</i> | NM_001005752 | AD | DFNA2B | |
| <i>GJB6</i> | <i>GJB6</i> | AR | DFNB82 | Includes regions regulating GJB2 Chudly-McCullough syndrome |
| <i>GPSM2</i> | <i>GPSM2</i> | AR | DFNB82 | |
| <i>GREB1L</i> | <i>GREB1L</i> | | | |
| <i>GRHL2</i> | NM_024915 | AD | DFNA28 | |
| <i>GRXCR1</i> | <i>GRXCR1</i> | AR | DFNB25 | |
| <i>GRXCR2</i> | <i>GRXCR2</i> | AR | DFNB101 | |
| <i>HARS</i> | <i>HARS</i> | AR | | Usher syndrome type 3B |
| <i>HARS2</i> | <i>HARS2</i> | AR | | Perrault syndrome 2 |
| <i>HGF</i> | <i>HGF</i> | AR | DFNB39 | |
| <i>HOMER2</i> | <i>HOMER2</i> | AD | | |
| <i>HSD17B4</i> | <i>HSD17B4</i> | AR | | Perrault syndrome 1 |
| <i>ILDR1</i> | <i>ILDR1</i> | AR | DFNB42 | |
| <i>KARS</i> | <i>KARS</i> | AR | DFNB89 | |
| <i>KCNE1</i> | <i>KCNE1</i> | AR | | |
| <i>KCNJ10</i> | <i>KCNJ10</i> | AR | | Jervell and Lange-Nielsen syndrome 2 |
| <i>KCNQ1</i> | <i>KCNQ1</i> | AD | | Pendred syndrome |
| <i>KCNQ4</i> | <i>KCNQ4</i> | AD | DFNA2A | Long QT syndrome 1 |
| <i>KITLG</i> | <i>KITLG</i> | AD | | |
| <i>LARS2</i> | <i>LARS2</i> | AR | | Waardenburg syndrom, type 2 |
| <i>LHFPL5</i> | <i>LHFPL5</i> | AR | DFNB66/67 | Perrault syndrome 4 |
| <i>LMX1A</i> | <i>LMX1A</i> | | | |
| <i>LOC653786</i> | | - | | OTOA Pseudogene |
| <i>LOXHD1</i> | <i>LOXHD1</i> | AR | DFNB77 | |
| <i>LOXL3</i> | <i>LOXL3</i> | AR | | possible Stickler syndrome |
| <i>LRTOMT</i> | <i>LRTOMT</i> | AR | DFNB63 | |
| <i>MARVELD2</i> | <i>MARVELD2</i> | AR | DFNB49 | |
| <i>MCM2</i> | <i>MCM2</i> | AD | | |
| <i>MET</i> | <i>MET</i> | AR | DFNB97 | |
| <i>MIRN96</i> | chr7:129,414,493-129,414,651 | AD | DFNA50 | |
| <i>MITF</i> | <i>MITF</i> | AD | | Waardenburg syndrome, type 2A |
| <i>MPZL2</i> | <i>MPZL2</i> | AR | DFNB111 | |
| <i>MSRB3</i> | <i>MSRB3</i> | AR | DFNB74 | |
| <i>MTRNR1</i> | chrM:650-1603 | M | | |
| <i>MTTE</i> | chrM:14675-14743 | M | | Maternally inherited diabetes and deafness (MIDD) |
| <i>MTTK</i> | chrM:8296-8365 | M | | MERRF, MTTK |
| <i>MTTL1</i> | chrM:3231-3305 | M | | MERRF syndrome |
| <i>MTTS1</i> | chrM:7447-7515 | M | | |
| <i>MYH14</i> | <i>MYH14</i> | AD | DFNA4 | |
| <i>MYH9</i> | <i>MYH9</i> | AD | DFNA17 | |
| <i>MYO15A</i> | <i>MYO15A</i> | AR | DFNB3 | |
| <i>MYO15A</i> | chr17:18026518-18026747 | AR | | MYO15A exon 1A |
| <i>MYO1A</i> | <i>MYO1A</i> | AD | DFNA48 ? | |
| <i>MYO3A</i> | <i>MYO3A</i> | AR | DFNB30 | |
| <i>MYO6</i> | <i>MYO6</i> | AR | DFNB37; DFNA22 | |
| <i>MYO7A</i> | <i>MYO7A</i> | AR,AD | DFNB2; DFNA11 | Usher syndrome, type 1B |
| <i>NARS2</i> | <i>NARS2</i> | AR | DFNB94 | |
| <i>NDP</i> | <i>NDP</i> | XLR | | Norrie disease |
| <i>NLRP3</i> | <i>NLRP3</i> | AD | | Muckle-Wells syndrome |
| <i>OPA1</i> | <i>OPA1</i> | AD | | Optic atrophy plus syndrome |
| <i>OSBPL2</i> | <i>OSBPL2</i> | AD | DFNA67 | |
| <i>OTOA</i> | <i>OTOA</i> | AR | DFNB22 | |
| <i>OTOF</i> | <i>OTOF</i> | AR | DFNB9 | |
| <i>OTOG</i> | <i>OTOG</i> | AR | DFNB18B | Auditory neuropathy |
| <i>OTOGL</i> | <i>OTOGL</i> | AR | DFNB84 | |
| <i>P2RX2</i> | <i>P2RX2</i> | AD | DFNA41 | |
| <i>PAX3</i> | <i>PAX3</i> | AD,AR | | Waardenburg syndrome, type 1 and type 3 |
| <i>PCDH15</i> | <i>PCDH15</i> | AR | | Usher syndrome, type 1F |
| <i>PDE1C</i> | <i>PDE1C</i> | AD | DFNA74 ? | |
| <i>PDZD7</i> | <i>PDZD7</i> | AR | | Usher syndrome, type IIC |
| <i>PEX1</i> | <i>PEX1</i> | AR | | Heimler syndrome |
| <i>PEX6</i> | <i>PEX6</i> | AR | | Heimler syndrome |
| <i>PJVK</i> | DFNB59 | AR | DFNB59 | Auditory neuropathy |
| <i>PNPT1</i> | <i>PNPT1</i> | AR | DFNB70 | |
| <i>POLR1C</i> | <i>POLR1C</i> | AR | | Treacher Collins syndrome 3 |
| <i>POLRID</i> | <i>POLRID</i> | AD | | Treacher Collins syndrome 2 |

| Gene (OMIM) | SureDesign input | Inherit | Locus (OMIM) | Syndrome and notes |
|--------------------|------------------|---------|--------------|-------------------------------|
| <i>POU3F4</i> | <i>POU3F4</i> | XR | DFNX2 | |
| <i>POU4F3</i> | <i>POU4F3</i> | AD | DFNA15 | |
| <i>PRPS1</i> | <i>PRPS1</i> | XR | DFNX1 | |
| <i>PTPRQ</i> | <i>PTPRQ</i> | AR | DFNB84 | |
| <i>PTRH2</i> | <i>PTRH2</i> | AR | | IMNEPD syndrome |
| <i>RDX</i> | <i>RDX</i> | AR | DFNB24 | |
| <i>ROR1</i> | <i>NM_005012</i> | AR | | |
| <i>SIPR2</i> | <i>SIPR2</i> | AR | DFNB68 | |
| <i>SEMA3E</i> | <i>SEMA3E</i> | AD | | CHARGE syndrome |
| <i>SERPINB6</i> | <i>SERPINB6</i> | AR | DFNB91 | |
| <i>SIX1</i> | <i>SIX1</i> | AD | DFNA23 | Branchio-oto-renal syndrome |
| <i>SIX5</i> | <i>SIX5</i> | | | Branchio-oto-renal syndrome |
| <i>SLC17A8</i> | <i>SLC17A8</i> | AD | DFNA25 | |
| <i>SLC22A4</i> | <i>SLC22A4</i> | AR | | |
| <i>SLC26A4</i> | <i>SLC26A4</i> | AR | | Pendred syndrome |
| <i>SLC26A5</i> | <i>SLC26A5</i> | AR | DFNB61 | |
| <i>SLITRK6</i> | <i>SLITRK6</i> | AR | | Deafness and myopia |
| <i>SMAC/DIABLO</i> | <i>DIABLO</i> | AD | DFNA64 | |
| <i>SMPX</i> | <i>SMPX</i> | XR | DFNX4 | |
| <i>SNAI2</i> | <i>SNAI2</i> | AR | | Waardenburg syndrome, type 2D |
| <i>SOX10</i> | <i>SOX10</i> | AD | | Waardenburg syndrome, type 4C |
| <i>STRC</i> | <i>STRC</i> | AR | DFNB16 | |
| <i>STRCP1</i> | | - | | STRC pseudogene |
| <i>SYNE4</i> | <i>SYNE4</i> | AR | DFNB76 | |
| <i>TBC1D24</i> | <i>TBC1D24</i> | AR,AD | DFNA65 | |
| <i>TBX1</i> | <i>TBX1</i> | AD | | Velocardiofacial syndrome |
| <i>TCOF1</i> | <i>TCOF1</i> | AD | | Treacher Collins syndrome 1 |
| <i>TECTA</i> | <i>TECTA</i> | AR,AD | DFNB21 | |
| <i>TECTB</i> | <i>TECTB</i> | | | mouse model |
| <i>TIMM8A</i> | <i>TIMM8A</i> | XR | | Mohr-Tranebjærg syndrome |
| <i>TJP2</i> | <i>TJP2</i> | AD | DFNA51 | |
| <i>TMC1</i> | <i>TMC1</i> | AR,AD | DFNB7/11 | |
| <i>TMEM132E</i> | <i>TMEM132E</i> | AR | DFNB99 | |
| <i>TMIE</i> | <i>TMIE</i> | AR | DFNB6 | |
| <i>TMPRSS3</i> | <i>TMPRSS3</i> | AR | DFNB8/10 | |
| <i>TNC</i> | <i>TNC</i> | AD | DFNA56 | |
| <i>TPRN</i> | <i>TPRN</i> | AR | DFNB79 | |
| <i>TRIOBP</i> | <i>TRIOBP</i> | AR | DFNB28 | |
| <i>TSPEAR</i> | <i>TSPEAR</i> | AR | DFNB98 | |
| <i>TWNK</i> | <i>NM_021830</i> | AR | | Perrault syndrome 5 |
| <i>USH1C</i> | <i>USH1C</i> | AR | DFNB18 | Usher syndrome, type 1C |
| <i>USH1G</i> | <i>USH1G</i> | AR | | Usher syndrome, type 1G |
| <i>USH2A</i> | <i>USH2A</i> | AR | | Usher syndrome, type 2A |
| <i>WBP2</i> | <i>WBP2</i> | AR | | |
| <i>WFS1</i> | <i>WFS1</i> | AD | DFNA6/14/38 | Wolfram syndrome |
| <i>WHRN</i> | <i>DFNB31</i> | AR | DFNB31 | Usher syndrome, type 2D |

Table S2A. Genes responsible for hearing loss in Palestinian families

| Gene | Number of families | Number of different mutations | Phenotype |
|-----------------|--------------------|-------------------------------|---------------------|
| <i>ADGRV1</i> | 4 | 3 | Usher 2C |
| <i>CACNA1D</i> | 4 | 1 | SANDD |
| <i>CDC14A</i> | 1 | 1 | DFNB32 |
| <i>CDH23</i> | 17 | 10 | DFNB12 / Usher 1D |
| <i>CLDN14</i> | 8 | 2 | DFNB29 |
| <i>CLPP</i> | 1 | 1 | Perrault syn |
| <i>COCH</i> | 1 | 1 | DFNA9 |
| <i>COL11A2</i> | 1 | 1 | DFNB53 |
| <i>COL4A6</i> | 1 | 1 | DFNX6 |
| <i>DFNA5</i> | 1 | 1 | DFNA5 |
| <i>EDNRB</i> | 2 | 2 | Waardenburg syn |
| <i>EPS8L2</i> | 2 | 2 | DFNB106 |
| <i>ESPN</i> | 2 | 2 | DFNB36 |
| <i>ESRRB</i> | 3 | 2 | DFNB35 |
| <i>GIPC3</i> | 1 | 1 | DFNB15 / JAMS |
| <i>GJB2</i> | 74 | 7 | DFNB1A |
| <i>GPSM2</i> | 5 | 4 | DFNB82 / CMCS |
| <i>HSD17B4</i> | 1 | 1 | Perrault syn |
| <i>ILDR1</i> | 3 | 2 | DFNB42 |
| <i>LARS2</i> | 4 | 1 | Perrault syn |
| <i>LHFPL5</i> | 2 | 2 | DFNB67 |
| <i>LOXHD1</i> | 2 | 2 | DFNB77 |
| <i>LRTOMT</i> | 1 | 1 | DFNB63 |
| <i>MARVELD2</i> | 1 | 1 | DFNB49 |
| <i>MYO3A</i> | 1 | 1 | DFNB30 |
| <i>MYO6</i> | 5 | 2 | DFNB37 / DFNA22 |
| <i>MYO7A</i> | 28 | 15 | DFNB2 / DFNA11 |
| <i>MYO15A</i> | 37 | 15 | DFNB3 |
| <i>OTOA</i> | 8 | 5 | DFNB22 |
| <i>OTOF</i> | 3 | 3 | AUNB1 |
| <i>PAX3</i> | 6 | 3 | Waardenburg syn |
| <i>PCDH15</i> | 7 | 2 | DFNB23 |
| <i>PJVK</i> | 2 | 1 | DFNB59 |
| <i>POU3F4</i> | 2 | 1 | DFNX2 |
| <i>POU4F3</i> | 2 | 2 | DFNA15 |
| <i>PTPRQ</i> | 3 | 2 | DFNB84A |
| <i>PTRH2</i> | 2 | 1 | IMNEPED |
| <i>RDX</i> | 1 | 1 | DFNB24 |
| <i>SLC26A4</i> | 30 | 14 | DFNB4 / Pendred syn |
| <i>STRC</i> | 1 | 1 | DFNB16 |
| <i>TBC1D24</i> | 1 | 1 | DFNB86 / DOORS |
| <i>TECTA</i> | 9 | 3 | DFNB21 |
| <i>TMC1</i> | 4 | 4 | DFNB7 |
| <i>TMPRSS3</i> | 15 | 6 | DFNB8/10 |
| <i>TRIOBP</i> | 21 | 4 | DFNB28 |
| <i>USH1C</i> | 2 | 1 | Usher 1C / DFNB18A |
| <i>USH1G</i> | 2 | 2 | Usher 1G |
| <i>USH2A</i> | 3 | 1 | Usher 2A |
| Total | 337 | 143 | |

Abbreviations:

AUNB1: Auditory neuropathy

CMCS: Chudley-McCullough syndrome

DOORS: Deafness, onychodystrophy, osteodystrophy, MR, and seizures

IMNEPED: Infantile-onset multisystem neurologic, endocrine, pancreatic disease

JAMS1: Juvenile audiogenic monogenic seizures

SANDD: Sinoatrial node dysfunction

Table S2B. Phenotypes and genotypes of all families with genetic diagnoses for their hearing loss

| Family | City | Consang | Single / multi HL | Aff in family | Inherit | Onset pre-post-lingual | Synd | Hearing loss | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position |
|---------------|-----------|---------|-------------------|---------------|---------|------------------------|------|--------------------------------|--------------------|-----------|----------------|-------------------------|------------------------------------|--------------------------------|
| CX | Ramallah | yes | multi | 2 | AR | pre | | moderate to severe | new | homoz | <i>ADGRV1</i> | c.2898G>A | p.(Glu966Glu) | chr5:89940686 |
| JO | Ramallah | yes | multi | 2 | AR | pre | | moderate | new | homoz | <i>ADGRV1</i> | c.2898G>A | p.(Glu966Glu) | chr5:89940686 |
| EI | Bethlehem | yes | multi | 3 | AR | pre | | moderate to severe | new | homoz | <i>ADGRV1</i> | c.10426 G>A | p.(Gly3476Arg) | chr5:90024750 |
| IJ | Bethlehem | yes | multi | 2 | AR | pre | yes | moderate to severe | new | homoz | <i>ADGRV1</i> | c.14315C>A | p.(Ser4772*) | chr5:90086961 |
| CT | Bethlehem | yes | multi | 4 | AR | pre | yes | moderate; cardiac signs | new | homoz | <i>CACNA1D</i> | c.1127C>T | p.(Ala376Val) | chr3:53707750 |
| HN | Jersualem | yes | sing | 1 | AR | pre | yes | moderate; prolonged AV conduce | new | homoz | <i>CACNA1D</i> | c.1127C>T | p.(Ala376Val) | chr3:53707750 |
| QS051-052 | Qalqilia | yes | sing | 1 | AR | pre | | | new | homoz | <i>CACNA1D</i> | c.1127C>T | p.(Ala376Val) | chr3:53707750 |
| X16-17 | Jersualem | yes | multi | 2 | AR | pre | | | new | homoz | <i>CACNA1D</i> | c.1127C>T | p.(Ala376Val) | chr3:53707750 |
| Q-N | Tulkarm | yes | multi | 4 | AR | pre | | severe | new | homoz | <i>CDC14A</i> | c.51-1G>T | p.(?) | chr1:100819317 |
| FO | Nablus | yes | multi | 2 | AR | pre | | profound | known | homoz | <i>CDH23</i> | c.683A>T | p.(Asp228Val) | chr10:73330605 |
| AS-KK | Jenin | yes | sing | 1 | AR | pre | | | known | homoz | <i>CDH23</i> | c.1036C>T | p.(Pro346Ser) | chr10:733707052 |
| DW | Ramallah | yes | multi | 2 | AR | pre | | severe to profound | known | homoz | <i>CDH23</i> | c.1036C>T | p.(Pro346Ser) | chr10:733707052 |
| G | Hebron | yes | multi | 5 | AR | pre | | severe to profound | known | homoz | <i>CDH23</i> | c.1036C>T | p.(Pro346Ser) | chr10:733707052 |
| DA (EP5-7) | Ramallah | yes | multi | 3 | AR | pre | | moderate | known | homoz | <i>CDH23</i> | c.1037C>T | p.(Pro346Ser) | chr10:733707053 |
| AA-N | Jenin | yes | multi | 4 | AR | pre | | severe to profound | known | homoz | <i>CDH23</i> | c.1675C>T | p.(Pro559Ser) | chr10:73437373 |
| GA-D | Gaza | yes | multi | 2 | AR | pre | | moderate | known | homoz | <i>CDH23</i> | c.1675C>T | p.(Pro559Ser) | chr10:73437373 |
| AB | Ramallah | no | multi | 6 | AR | pre | | moderate | known | cpd het | <i>CDH23</i> | c.[(1675C>T);(3211G>A)] | p.(Pro559Ser);(Glu1071Lys) | chr10:73437373; chr10:73468959 |
| EE | Ramallah | yes | multi | 2 | AR | pre | | moderate | known | homoz | <i>CDH23</i> | c.3211G>A | p.(Glu1071Lys) | chr10:73468959 |
| HS8 | Ramallah | yes | sing | 1 | AR | pre | | | known | homoz | <i>CDH23</i> | c.3211G>A | p.(Glu1071Lys) | chr10:73468959 |
| EG | Hebron | yes | multi | 3 | AR | pre | yes | profound | new | homoz | <i>CDH23</i> | c.4562A>G | p.(Asn1521Ser) | chr10:73500652 |
| EPIC | Bethlehem | yes | multi | 3 | AR | pre | yes | profound, RP, epilepsy | new | homoz | <i>CDH23</i> | c.5749G>A | p.(Glu1917Lys) | chr10:73545424 |
| A151 | Jersualem | yes | sing | 1 | AR | pre | | moderate | known | homoz | <i>CDH23</i> | c.6614C>T | p.(Pro205Leu) | chr10:73553299 |
| IP | Nablus | yes | sing | 1 | AR | pre | | moderate to severe | known | homoz | <i>CDH23</i> | c.6614C>T | p.(Pro205Leu) | chr10:73553299 |
| DF (EP10-12) | Bethlehem | yes | multi | 2 | AR | pre | | | new | homoz | <i>CDH23</i> | c.8140G>T | p.(Asp2714Tyr) | chr10:73566000 |
| BV3 (EP23-24) | Jersualem | yes | multi | 3 | AR | pre | | profound | new | homoz | <i>CDH23</i> | c.8204T>C | p.(Leu2735Pro) | chr10:73567059 |
| EP25 | Bethlehem | yes | sing | 1 | AR | pre | | severe to profound | new | homoz | <i>CDH23</i> | c.8204T>C | p.(Leu2735Pro) | chr10:73567059 |
| X13 | Jersualem | yes | sing | 1 | AR | pre | | | new | homoz | <i>CLDN14</i> | c.664delG | p.(Ala22Prof8*) | chr21:37833330 |
| AA-G | Jenin | yes | multi | 3 | AR | pre | | severe to profound | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| AA-K | Jenin | yes | multi | 3 | AR | pre | | severe to profound | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| AA-O | Jenin | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| AS-X | Jenin | yes | multi | 5 | AR | pre | | | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| DB | Hebron | yes | multi | 3 | AR | pre | | severe to profound | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| QS039 | Qalqilia | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| FN | Jenin | yes | multi | 3 | AR | pre | | profound w/mild MR, DD | known | homoz | <i>CLDN14</i> | c.83C>T | p.(Pro28Leu) | chr21:37833911 |
| GA-H | Gaza | yes | multi | 3 | AR | pre | | severe to profound | new | homoz | <i>CLPP</i> | c.270+3G>T | p.(?) | chr19:6361954 |
| CO | Jenin | no | multi | 9 | AD | post | | progressive, onset 20-30y | new | het | <i>COCH</i> | c.1625G>C | p.(Cys542Ser) | chr14:31358969 |
| Q-U | Tulkarm | yes | multi | 4 | AR | pre | yes | severe with joint problems | new | homoz | <i>COL1A2</i> | c.3080G>A | p.(Gly1027Glu) | chr6:33139560 |
| AS-CC | Jenin | yes | sing | 1 | XR | pre | | any cochlear malformation? | new | hemiz | <i>COL4A6</i> | c.1549C>T | p.(Arg517*) | chrX:107431785 |
| BL | Nablus | no | multi | 5 | AD | post | | progressive, onset ~17y | known | het | <i>DFNA5</i> | c.991-15_991-13del | p.(?) | chr7:24746008 |
| Q-R | Tulkarm | yes | sing | 1 | AD | pre | yes | severe w/ Waardenberg | new | het | <i>EDNRB</i> | c.1071+2T>C | p.(?) | chr13:78477289 |
| JS | Tulkarm | yes | sing | 1 | AR | pre | yes | Waardenberg syndrome | new | homoz | <i>EDNRB</i> | c.823G>A | p.(Val275Met) | chr13:78477673 |
| IW | Nablus | yes | multi | 3 | AR | pre | yes | profound | new | homoz | <i>EPS8L2</i> | c.148insGACA | p.(Ser50Trpfs*34) | chr11:710469 |
| GX | Nablus | yes | multi | 4 | AR | post | | progressive, onset ~6y | new | homoz | <i>EPS8L2</i> | c.1430dup | p.(Val478Serfs*25) | chr11:723327 |
| AA-E | Jenin | yes | sing | 1 | AR | pre | | profound | new | homoz | <i>ESPN</i> | c.2081_2082del | p.(Ser694Cysfs*51) | chr1:65119111 |
| Q-S | Tulkarm | yes | sing | 1 | AD | post | | progressive, moderate | known | het | <i>ESPN</i> | c.2155A>C | p.(Ser719Arg) | chr1:6511986 |
| BP | Jersualem | yes | multi | 3 | AR | pre | | severe to profound | new | homoz | <i>ESRRB</i> | c.545G>A | p.(Arg182His) | chr14:76948389 |
| AS-S | Jenin | yes | multi | 2 | AR | pre | | | new | homoz | <i>ESRRB</i> | c.787G>A | p.(Gly263Ser) | chr14:76949102 |
| FT | Jenin | yes | multi | 3 | AR | pre | | profound | new | homoz | <i>ESRRB</i> | c.787G>A | p.(Gly263Ser) | chr14:76949102 |
| AA-F | Jenin | yes | multi | 2 | AR | pre | yes | severe to profound w/ epilepsy | known | homoz | <i>GIPC3</i> | c.122C>A | p.(Thr41Lys) | chr19:3585717 |
| AS-C | Jenin | yes | sing | 1 | AR | pre | | | known | homoz | <i>GJB2</i> | c.358_360delGAG | p.(Glu120del) | chr13:20763361 |
| GV | Jenin | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.358_360delGAG | p.(Glu120del) | chr13:20763361 |
| DG | Jersualem | yes | multi | 3 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.235delC | p.(Leu79Cysfs*3) | chr13:20763486 |
| FM | Hebron | yes | multi | 2 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.235delC | p.(Leu79Cysfs*3) | chr13:20763486 |
| IZ | Hebron | yes | sing | 1 | AR | pre | | moderate to severe | known | homoz | <i>GJB2</i> | c.235delC | p.(Leu79Cysfs*3) | chr13:20763486 |
| X33 | Jersualem | yes | sing | 1 | AR | pre | | | known | homoz | <i>GJB2</i> | c.235delC | p.(Leu79Cysfs*3) | chr13:20763486 |
| X18 | Jersualem | no | sing | 1 | AR | pre | | | known | cpd het | <i>GJB2</i> | c.[(235delC);(290insA)] | p.[(Leu79Cysfs*3);(Arg98Thrf8*15)] | chr13:20763486; chr10:20763430 |
| X23 | Jersualem | no | sing | 1 | AR | pre | | | known | cpd het | <i>GJB2</i> | c.[(235delC);(290insA)] | p.[(Leu79Cysfs*3);(Arg98Thrf8*15)] | chr13:20763486; chr10:20763430 |
| B6-7 | Hebron | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.229T>C | p.(Trp77Arg) | chr13:20763492 |
| AF | Hebron | yes | multi | 3 | AR | pre | | | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |
| L | Hebron | yes | multi | 6 | AR | pre | | | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |
| S | Hebron | yes | multi | 17 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |
| X6 | Hebron | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |
| a16 | Hebron | yes | sing | 1 | AR | pre | | severe to profound | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |

| Family | City | Consang | Single / multi | Aff in family | Onset pre-post-lingual | Synd | Hearing loss | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position |
|-----------|-----------|---------|----------------|---------------|------------------------|------|-------------------------|--------------------|-----------|----------------|---------------------------|------------------------------------|--------------------------------|
| a31 | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.167delT | p.(Leu56Argfs*26) | chr13:20763554 |
| GI | Hebron | yes | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(167delT);(c.235delC)] | p.(Leu56Argfs*26); (Leu79Cysfs*3)] | chr13:20763554; chr10:20763486 |
| D | Hebron | yes | multi | 8 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(167delT);(c.235delC)] | p.(Leu56Argfs*26); (Leu79Cysfs*3)] | chr13:20763554; chr10:20763486 |
| A127-129 | Jersualem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-C | Jenin | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-I | Jenin | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-J | Jenin | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-M | Jenin | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-P | Jenin | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-R | Jenin | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-Y | Jenin | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AA-Z | Jenin | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AC | Hebron | yes | sing | 1 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AQ | Tulkarem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS | Hebron | yes | multi | 2 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS-D | Jenin | yes | sing | 1 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS-DD | Jenin | yes | multi | 4 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS-G | Jenin | yes | sing | 1 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS-U | Jenin | yes | multi | 2 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| AS-Y | Jenin | yes | sing | 1 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| B3-5 | Hebron | yes | multi | 2 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| BN | Bethlehem | yes | multi | 4 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| BO | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| BS | Bethlehem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| CE | Jenin | yes | multi | 5 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| CP | Hebron | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| CW | Jenin | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| DM | Hebron | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| EP1-4 | Bethlehem | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| FI | Jersualem | no | multi | 3 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| FJ | Jersualem | yes | multi | 6 | AR | pre | severe | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| FR | Hebron | yes | multi | 5 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| FS | Salfleet | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| GA-L | Gaza | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| GC | Hebron | yes | multi | 6 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| GG | Qalqilia | yes | multi | 2 | AR | pre | moderate | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| GK | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| HE | Nablus | no | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| HO | Jenin | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| HS1 | Ramallah | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| HS4 | Ramallah | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| IN | Nablus | yes | sing | 1 | AR | pre | profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| JD | Ramallah | yes | multi | 2 | AR | pre | moderate | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| JG | Bethlehem | yes | sing | 1 | AR | pre | moderate | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| JI | Bethlehem | yes | multi | 3 | AR | pre | moderate to severe | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| Q-A | Tulkarm | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| Q-E | Tulkarm | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| Q-M | Tulkarm | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| QS036-037 | Qalqilia | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| X29 | Jersualem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| a36 | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GJB2</i> | c.35delG | p.(Gly12Valfs*2) | chr13:20763686 |
| FP | Ramallah | no | sing | 1 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.235delC)] | p.[(Gly12Valfs*2);(Leu79Cysfs*3)] | chr13:20763686; chr13:20763486 |
| HV | Hebron | yes | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.235delC)] | p.[(Gly12Valfs*2);(Leu79Cysfs*3)] | chr13:20763686; chr13:20763486 |
| J1-2 | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.235delC)] | p.[(Gly12Valfs*2);(Leu79Cysfs*3)] | chr13:20763686; chr13:20763486 |
| X37-38 | Jersualem | no | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.235delC)] | p.[(Gly12Valfs*2);(Leu79Cysfs*3)] | chr13:20763686; chr13:20763486 |
| GO | Jenin | no | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.-23+1G>A)] | p.[(Gly12Valfs*2);(?)] | chr13:20763686; chr13:20766921 |
| IK | Bethlehem | no | multi | 2 | AR | pre | moderate to severe | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.-23+1G>A)] | p.[(Gly12Valfs*2);(?)] | chr13:20763686; chr13:20766921 |
| J3-5 | Hebron | yes | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>GJB2</i> | c.[(35delG);(c.-23+1G>A)] | p.[(Gly12Valfs*2);(?)] | chr13:20763686; chr13:20766921 |
| X12 | Jersualem | yes | sing | 1 | AR | pre | severe | known | homoz | <i>GPSM2</i> | c.832C>T | p.(Arg278*) | chr1:109444446 |
| GZ | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GPSM2</i> | c.977G>A | p.(Trp326*) | chr1:109445771 |
| GA-N | Gaza | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>GPSM2</i> | c.977G>A | p.(Trp326*) | chr1:109445771 |
| a33 | Hebron | yes | multi | 6 | AR | pre | severe to profound | known | homoz | <i>GPSM2</i> | c.1492C>T | p.(Arg498*) | chr1:109465090 |
| CC | Bethlehem | yes | sing | 1 | AR | pre | severe to profound w/ID | known | homoz | <i>GPSM2</i> | c.379C>T | p.(Arg127*) | chr1:109440214 |
| CG | Qalqilia | yes | multi | 13 | AR | pre | severe to profound | new | homoz | <i>HSD17B4</i> | c.1842G>A at splice | p.(?) | chr5:118862914 |
| EY | Nablus | yes | sing | 1 | AR | pre | severe to profound | | | | | | |

| Family | City | Consang | Single / multi | Aff in HL | family | Inherit | Onset pre-post-lingual | Synd | Hearing loss | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position |
|------------------------------|-----------|---------|----------------|-----------|--------|---------|--------------------------------|-----------------------------|--------------|--------------------|-------------|-----------------|--------------------------------|-------------------------------------|--------------------------------|
| AS-BB | Jenin | yes | multi | 4 | AR | pre | severe to profound | | | new | homoz | <i>ILDR1</i> | c.820C>T | p.(Gln274*) | chr3:121712776 |
| II | Jenin | yes | multi | 3 | AR | pre | severe to profound | | | new | homoz | <i>ILDR1</i> | c.C820T | p.(Gln274*) | chr3:121712776 |
| GA-O | Gaza | yes | multi | 2 | AR | pre | profound | | | new | homoz | <i>ILDR1</i> | c.294delA | p.(Val99Phefs*3) | chr3:121724176 |
| BB | Bethlehem | yes | multi | 3 | AR | pre | severe | | | new | homoz | <i>LARS2</i> | c.457A>C | p.(Asn153His) | chr3:45461162 |
| GA-A | Gaza | yes | multi | 2 | AR | pre | profound | | | new | homoz | <i>LARS2</i> | c.457A>C | p.(Asn153His) | chr3:45461162 |
| HS | Gaza | yes | multi | 2 | AR | pre | profound | | | new | homoz | <i>LARS2</i> | c.457A>C | p.(Asn153His) | chr3:45461162 |
| Q-F | Tulkarm | yes | multi | 2 | AR | pre | yes | severe to profound | | new | homoz | <i>LARS2</i> | c.457A>C | p.(Asn153His) | chr3:45461162 |
| DD | Bethlehem | yes | multi | 5 | AR | pre | severe to profound | | | known | homoz | <i>LHFPL5</i> | c.1A>G | p.(Met1Val) | chr6:35773448 |
| CK | Salfleet | yes | multi | 1 | AR | pre | severe to profound | | | new | homoz | <i>LHFPL5</i> | c.676+1G>A | p.(?) | chr6:35787241 |
| AH | Bethlehem | yes | multi | 6 | AR | pre | profound | | | new | homoz | <i>LOXHD1</i> | del ex 22-25 out of frame | p.(?) | chr18:44118336_44136525 |
| QS044-047 | Qalqilia | no | multi | 2 | AR | pre | profound | | | new | cpd het | <i>LOXHD1</i> | c.[(3404_3407dup);(c.4843G>C)] | p.[(Arg1137Valfs*9);(Gly1615Arg)] | chr18:44126967; chr18:44104462 |
| GA-R | Gaza | yes | multi | 3 | AR | pre | profound | | | new | homoz | <i>LRTOMT</i> | c.327C>T | p.(Cys109Cys) | chr11:71817225 |
| X39 | Jersualem | yes | sing | 1 | AR | pre | | | | new | homoz | <i>MARVELD2</i> | c.608dupT | p.(Leu203Phefs*18) | chr5:68715820 |
| BA | Hebron | yes | multi | 7 | AR | pre | profound | | | new | cpd het | <i>MYO15A</i> | c.[(1223C>T);(4240G>A)] | p.[(Ala408Val);(Glu1414Lys)] | chr17:18023337; chr17:18035800 |
| C | Hebron | yes | multi | 11 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.3609+985G>A | p.(?) | chr17:18026708 |
| FG | Hebron | yes | multi | 4 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.3609+985G>A | p.(?) | chr17:18026708 |
| GJ | Hebron | yes | multi | 8 | AR | pre | severe | | | new | homoz | <i>MYO15A</i> | c.3609+985G>A | p.(?) | chr17:18026708 |
| GL | Hebron | yes | multi | 4 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.3609+985G>A | p.(?) | chr17:18026708 |
| HG | Hebron | yes | multi | 2 | AR | pre | moderate mixed | | | new | homoz | <i>MYO15A</i> | c.3609+985G>A | p.(?) | chr17:18026708 |
| IG | Hebron | yes | multi | 2 | AR | pre | moderate | | | new | cpd het | <i>MYO15A</i> | c.[(3609+985G>A);(7207G>T)] | p.([(?);(Asp2403Tyr)] | chr17:18026708; chr17:18052889 |
| E | Hebron | no | multi | 5 | AR | pre | moderate to severe | | | new | cpd het | <i>MYO15A</i> | c.[(3609+985G>A);(9572G>A)] | p.([(?);(Arg3191His)] | chr17:18026708; chr17:18065953 |
| AR | Hebron | no | sing | 1 | AR | pre | | | | new | cpd het | <i>MYO15A</i> | c.[(3609+985G>A);(9572G>A)] | p.([(?);(Arg3191His)] | chr17:18026708; chr17:18065953 |
| EP28 | Bethlehem | yes | sing | 1 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.3866+1G>A | p.(?) | chr17:18029771 |
| IO | Salfleet | yes | multi | 2 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.4142+5G>A | p.(?) | chr17:18034461 |
| QS027 | Qalqilia | yes | multi | 3 | AR | pre | | | | new | homoz | <i>MYO15A</i> | c.4142+5G>A | p.(?) | chr17:18034661 |
| CI | Hebron | yes | sing | 1 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.4240G>A | p.(Glu1414Lys) | chr17:18035800 |
| DC ($\alpha 15-\alpha 25$) | Hebron | yes | multi | 3 | AR | pre | profound | | | new | homoz | <i>MYO15A</i> | c.4240G>A | p.(Glu1414Lys) | chr17:18035800 |
| X3 | Jersualem | yes | sing | 1 | AR | pre | | | | known | homoz | <i>MYO15A</i> | c.6340G>A | p.(Val114Met) | chr17:18049252 |
| X24 | Jersualem | no | sing | 1 | AR | pre | | | | new | cpd het | <i>MYO15A</i> | c.[(6499T>C);(8767C>T)] | p.([Trp2167His];(Arg2923*)) | chr17:18049411; chr17:18060523 |
| IE | Hebron | yes | multi | 4 | AR | pre | HL and cleft palate (IE3 only) | | | known | homoz | <i>MYO15A</i> | c.6893G>A | p.(Arg298Gln) | chr17:18052203 |
| AL | Hebron | yes | multi | 3 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| AN | Hebron | yes | multi | 4 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| BW | Hebron | yes | multi | 5 | AR | pre | moderate to severe | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| CF | Hebron | yes | multi | 2 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| DX | Bethlehem | yes | multi | 3 | AR | pre | severe | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| HH | Hebron | yes | sing | 1 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| HI | Bethlehem | yes | multi | 2 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| M | Hebron | yes | multi | 2 | AR | pre | profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| P | Hebron | yes | multi | 9 | AR | pre | | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| $\alpha 18$ | Hebron | yes | sing | 1 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| $\alpha 19-\alpha 20$ | Hebron | yes | multi | 2 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| $\alpha 8-\alpha 9$ | Hebron | yes | multi | 3 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 |
| GP | Hebron | no | sing | 1 | AR | pre | | | | new | cpd het | <i>MYO15A</i> | c.[(7207G>T);(8183G>A)] | p.([(Asp2403Tyr);(Arg2728His)]) | chr17:18052889; chr17:18058028 |
| FY | Hebron | yes | multi | 2 | AR | pre | | | | known | homoz | <i>MYO15A</i> | c.8183G>A | p.(Arg2728His) | chr17:18058028 |
| HJ | Hebron | yes | sing | 1 | AR | pre | severe to profound | | | known | homoz | <i>MYO15A</i> | c.8183G>A | p.(Arg2728His) | chr17:18058028 |
| Z | Hebron | yes | multi | 3 | AR | pre | | | | known | homoz | <i>MYO15A</i> | c.8183G>A | p.(Arg2728His) | chr17:18058028 |
| BZ | Bethlehem | yes | multi | 8 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.8309_8311del | p.(Glu2770del) | chr17:18058506 |
| JQ | Tubas | yes | sing | 1 | AR | pre | severe to profound | | | new | homoz | <i>MYO15A</i> | c.8788+5G>T | p.(?) | chr17:18060549 |
| AI | Ramallah | yes | multi | 3 | AR | pre | moderate to severe | | | new | homoz | <i>MYO15A</i> | c.9572G>A | p.(Arg3191His) | chr17:18065953 |
| CQ | Ramallah | yes | multi | 2 | AR | pre | severe to profound | | | new | cpd het | <i>MYO15A</i> | c.[(9572G>A);(8309_8311del)] | p.([(Arg3191His);(Glu2770del)]) | chr17:18065953; chr17:18058506 |
| A132 | Jersualem | yes | sing | 1 | AR | pre | | | | check | homoz | <i>MYO3A</i> | c.1370_1371del | p.(Arg457Asnfs*25) | chr10:26377142 |
| AV | Nablus | yes | multi | 6 | AR | post | profound | | | known | homoz | <i>MYO6</i> | c.897G>T | p.(Glu299Asp) | chr6:76554694 |
| X36 | Jersualem | yes | sing | 1 | AR | pre | | | | homoz | <i>MYO6</i> | c.897G>T | p.(Glu299Asp) | chr6:76554694 | |
| AS-V | Jenin | yes | sing | 1 | AR | pre | | | | known | homoz | <i>MYO6</i> | c.2777T>A | p.(Leu926Gln) | chr6:76599892 |
| AS-W | Jenin | yes | multi | 5 | AR | pre | | | | known | homoz | <i>MYO6</i> | c.2777T>A | p.(Leu926Gln) | chr6:76599892 |
| QS009 | Qalqilia | yes | multi | 3 | AR | pre | profound | | | known | homoz | <i>MYO6</i> | c.2777T>A | p.(Leu926Gln) | chr6:76599892 |
| AS-R | Jenin | yes | sing | 1 | AR | pre | | | | new | homoz | <i>MYO7A</i> | c.4951G>A | p.(Gly1651Ser) | chr11:72912591 |
| X5 | Jersualem | yes | sing | 1 | AR | pre | | | | known | homoz | <i>MYO7A</i> | c.700C>T | p.(Gln234*) | chr11:76868015 |
| QS017 | Qalqilia | yes | multi | 8 | AR | pre | severe to profound | | | known | homoz | <i>MYO7A</i> | c.1117C>T | p.(Arg373Cys) | chr11:76871245 |
| JH | Salfleet | no | multi | 3 | AR | pre | moderate to severe, w/ Usher | yes | | known | cpd het | <i>MYO7A</i> | c.[(1583T>G);(6229dup)] | p.([(Leu528Pro);(Trp2077Leufs*51)]) | chr11:76873927; chr11:76922374 |
| X40 | Jersualem | yes | sing | 1 | AR | pre | | | | known | homoz | <i>MYO7A</i> | c.2187+1G>T | p.(?) | chr11:76886511 |
| GU | Jenin | yes | multi | 2 | AR | pre | yes | severe to profound, balance | | known | homoz | <i>MYO7A</i> | c.2307del | p.(Asn769Lysfs*5) | chr11:76890115 |
| AA-A | Jenin | yes | multi | 3 | AR | pre | yes | severe to profound | | new | homoz | <i>MYO7A</i> | c.2683C>T | p.(Arg895Cys) | chr11:76891516 |
| X19 | Jersualem | yes | sing | 1 | AR | pre | | | | known | homoz | <i>MYO7A</i> | c.3892G>A | p.(Gly1298Arg) | chr11:76901883 |
| QS004 | Qalqilia | yes | multi | 3 | AR | pre | profound | | | known | homoz | <i>MYO7A</i> | c.4153-2A>G | p.(?) | chr11:76905397 |
| QS013 | Qalqilia | yes | multi | 2 | AR | pre | profound | | | known | homoz | <i>MYO7A</i> | c.4153-2A>G | p.(?) | chr11:76905397 |

| Family | City | Consang | Single / multi | Aff in family | Onset pre-post-lingual | Synd | Hearing loss | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position | |
|-----------|-----------|---------|----------------|---------------|------------------------|------|-------------------------------|------------------------|-----------|----------------|-------------------------------|--------------------------|-----------------------------------|----------------|
| QS042-043 | Qalqilia | yes | sing | 1 | AR | pre | | known | homoz | <i>MYO7A</i> | c.4153-2A>G | p.(?) | chr11:76905397 | |
| AP | Hebron | yes | sing | 1 | AR | pre | severe to profound | new | homoz | <i>MYO7A</i> | c.4297C>T | p.(Gln1433*) | chr11:76905543 | |
| IT | Jersualem | no | multi | 6 | AR | pre | profound | known | cpd het | <i>MYO7A</i> | c.[(5095C>T);(700C>T)] | p.[(Gln1699*);(Gln234*)] | chr11:76913396; chr11:76868015 | |
| Q-Q | Tulkarm | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.5101C>T | p.(Arg1701*) | chr11:76913402 | |
| AO | Hebron | yes | multi | 5 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6196C>T | p.(Gln2066*) | chr11:76922341 | |
| BV6 | Jersualem | yes | multi | 3 | AR | pre | profound | known | homoz | <i>MYO7A</i> | c.6196C>T | p.(Gln2066*) | chr11:76922341 | |
| DT | Hebron | yes | multi | 2 | AR | pre | | known | homoz | <i>MYO7A</i> | c.6196C>T | p.(Gln2066*) | chr11:76922341 | |
| GB | Hebron | yes | multi | 3 | AR | pre | | known | homoz | <i>MYO7A</i> | c.6196C>T | p.(Gln2066*) | chr11:76922341 | |
| α12 | Hebron | yes | multi | 3 | AR | pre | | known | homoz | <i>MYO7A</i> | c.6196C>T | p.(Gln2066*) | chr11:76922341 | |
| DI | Nablus | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6211C>T | p.(Gln2071*) | chr11:76922356 | |
| HY | Ramallah | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6211C>T | p.(Gln2071*) | chr11:76922356 | |
| QS025-026 | Qalqilia | yes | sing | 1 | AR | pre | profound | known | homoz | <i>MYO7A</i> | c.6211C>T | p.(Gln2071*) | chr11:76922356 | |
| A118-119 | Jersualem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| FD | Ramallah | yes | sing | 1 | AR | pre | moderate | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| J9-17 | Hebron | yes | multi | 7 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| QS014-015 | Qalqilia | yes | multi | 2 | AR | pre | | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| QS023-024 | Qalqilia | yes | sing | 1 | AR | pre | | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| X31 | Jersualem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>MYO7A</i> | c.6487G>A | p.(Gly2163Ser) | chr11:76924953 | |
| DO | Hebron | yes | multi | 5 | AR | pre | severe to profound | new | homoz | <i>OTOA</i> | del 320-550 kb, complete gene | | chr16:21675289_21772050 or larger | |
| GQ | Hebron | yes | multi | 7 | AR | pre | moderate | new | homoz | <i>OTOA</i> | del 162-373 kb, complete gene | | chr16:21675289_21772050 or larger | |
| X10 | Jersualem | yes | multi | 2 | AR | pre | | known | homoz | <i>OTOA</i> | del >86kb, complete gene | | chr16:21687467_21773938 or larger | |
| X30 | Jersualem | yes | multi | 2 | AR | pre | | known | homoz | <i>OTOA</i> | del >86kb, complete gene | | chr16:21687467_21773938 or larger | |
| BR | Bethlehem | yes | multi | 7 | AR | pre | severe to profound | new | homoz | <i>OTOA</i> | c.1025A>T | p.(Asp342Val) | chr16:21716534 | |
| HT | Hebron | yes | sing | 1 | AR | pre | moderate to severe | new | homoz | <i>OTOA</i> | c.1025A>T | p.(Asp342Val) | chr16:21716534 | |
| X1 | Hebron | yes | sing | 1 | AR | pre | | new | homoz | <i>OTOA</i> | c.2017-2A>G | p.(?) | chr16:21739560 | |
| X8-9 | Jersualem | yes | multi | 2 | AR | pre | | new | homoz | <i>OTOA</i> | c.2017-2A>G | p.(?) | chr16:21739560 | |
| AM | Bethlehem | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>OTOF</i> | c.4030C>T | p.(Arg344*) | chr2:26691336 | |
| IM | Jersualem | yes | multi | 2 | AR | pre | profound | known | homoz | <i>OTOF</i> | c.2239G>T | p.(Glu747*) | chr2:26700593 | |
| FH | Hebron | yes | multi | 3 | AR | pre | profound | new | homoz | <i>OTOF</i> | c.1477C>T | p.(Arg1583Cys) | chr2:26688592 | |
| DL | Bethlehem | yes | multi | 2 | AD | pre | yes | Waardenberg syn type I | known | het | <i>PAX3</i> | c.812G>A | p.(Arg271His) | chr2:223086087 |
| BK | Jersualem | yes | multi | 2 | AD | pre | yes | Waardenburg syn type I | known | het | <i>PAX3</i> | c.251C>T | p.(Ser84Phe) | chr2:223161767 |
| DH | Hebron | yes | multi | 14 | AD | pre | yes | Waardenburg syn type I | known | het | <i>PAX3</i> | c.251C>T | p.(Ser84Phe) | chr2:223161767 |
| EP26 | Bethlehem | no | multi | 2 | AD | pre | yes | Waardenburg syn type I | known | het | <i>PAX3</i> | c.251C>T | p.(Ser84Phe) | chr2:223161767 |
| EP41 | Bethlehem | no | sing | 1 | AD | pre | yes | Waardenburg syn type I | known | het | <i>PAX3</i> | c.218C>T | p.(Ser84Phe) | chr2:223161800 |
| JM | Hebron | yes | sing | 2 | AD | pre | yes | Waardenburg syn type I | new | het | <i>PAX3</i> | c.202C>T | p.(Arg68Trp) | chr2:223161816 |
| A161 | Jersualem | yes | sing | 1 | AR | pre | moderate | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| AS-T | Jenin | yes | multi | 3 | AR | pre | | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| CA | Salfleet | yes | sing | 1 | AR | pre | severe to profound | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| EX | Qalqilia | yes | multi | 3 | AR | pre | severe to profound | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| GY | Salfleet | yes | multi | 3 | AR | pre | severe to profound | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| X21 | Jersualem | yes | sing | 1 | AR | pre | | new | homoz | <i>PCDH15</i> | c.4726C>T | p.(Gln1576*) | chr10:55569099 | |
| GA-U | Gaza | yes | multi | 2 | AR | pre | | new | homoz | <i>PCDH15</i> | c.432C>G | p.(Asp144Glu) | chr10:56128937 | |
| A | Hebron | yes | multi | 12 | AR | pre | severe to profound | known | homoz | <i>PJVK</i> | c.406C>T | p.(Arg136*) | chr2:179319253 | |
| B1-2 | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>PJVK</i> | c.406C>T | p.(Arg136*) | chr2:179319253 | |
| GW | Nablus | yes | multi | 6 | XR | pre | severe to profound | known | hemiz | <i>POU3F4</i> | c.G845T | p.(Arg282Leu) | chrX:82764177 | |
| JE | Tubas | no | multi | 5 | XR | pre | moderate | known | hemiz | <i>POU3F4</i> | c.G845T | p.(Arg282Leu) | chrX:82764177 | |
| GA-G | Gaza | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>POU4F3</i> | c.54deIA | p.(Glu18Aspfs*66) | chr5:145718729 | |
| X32 | Jersualem | yes | sing | 1 | AR | pre | | new | homoz | <i>POU4F3</i> | c.694G>T | p.(Glu232*) | chr5:145719684 | |
| CN | Bethlehem | yes | multi | 3 | AR | pre | moderate | known | homoz | <i>PTPRQ</i> | c.769G>T | p.(Glu257*) | chr12:80878248 | |
| DP | Bethlehem | yes | multi | 3 | AR | pre | moderate, vestibular problems | known | homoz | <i>PTPRQ</i> | c.769G>T | p.(Glu257*) | chr12:80878248 | |
| AS-EE | Jenin | yes | multi | 3 | AR | pre | severe to profound | new | homoz | <i>PTPRQ</i> | c.6730C>T | p.(Gln2244*) | chr12:81067075 | |
| EC | Hebron | yes | multi | 2 | AR | pre | yes | severe w/mild MR, DD | known | homoz | <i>PTRH2</i> | c.254A>C | p.(Gln85Pro) | chr17:57775086 |
| FF | Hebron | yes | multi | 2 | AR | pre | yes | severe w/mild MR, DD | known | homoz | <i>PTRH2</i> | c.254A>C | p.(Gln85Pro) | chr17:57775086 |
| IH | Hebron | yes | sing | 1 | AR | pre | severe to profound | new | homoz | <i>RDX</i> | c.513_514delAA | p.(Arg171Serfs*) | chr11:110128868 | |
| AS-P | Jenin | yes | sing | 1 | AR | pre | severe to profound | new | homoz | <i>SLC26A4</i> | c.284G>A | p.(Gly95Glu) | chr7:107303860 | |
| GA-Q | Gaza | yes | multi | 2 | AR | pre | severe to profound | new | homoz | <i>SLC26A4</i> | c.304G>A | p.(Gly102Arg) | chr7:107303880 | |
| JF1 | Ramallah | yes | multi | 4 | AR | pre | moderate to severe | new | homoz | <i>SLC26A4</i> | c.304G>A | p.(Gly102Arg) | chr7:107303880 | |
| EZ | Nablus | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.397T>A | p.(Ser133Thr) | chr7:107312675 | |
| Y | Hebron | yes | multi | 10 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.716T>A | p.(Val239Asp) | chr7:107315505 | |
| AA-L | Jenin | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| BF | Bethlehem | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| C5/C8 | Hebron | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| DY | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| EP13-14 | Bethlehem | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| EP35 | Bethlehem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |
| HQ | Gaza | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 | |

| Family | City | Consang | Single / multi | Aff in family | Onset pre-post-lingual | Synd | Hearing loss | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position |
|--------------------------------|-----------|---------|----------------|---------------|------------------------|------|------------------------------|--------------------|-----------|----------------|--------------------------------|-----------------------------------|--------------------------------|
| JF2 | Ramallah | yes | sing | 1 | AR | pre | moderate to severe | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 |
| X4 | Bethlehem | yes | multi | 5 | AR | pre | profound | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 |
| a22 | Hebron | yes | sing | 1 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.1001G>T | p.(Gly334Val) | chr7:107323982 |
| EO | Ramallah | yes | sing | 1 | AR | pre | | new | cpd het | <i>SLC26A4</i> | c.[(1054delG);(334C>T)] | p.[(Ala352Hifs*16);(Pro112Ser)] | chr7:107329550; chr7:107312612 |
| EM | Hebron | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1061T>C | p.(Phe354Ser) | chr7:107329557 |
| CS | Jenin | yes | multi | 6 | AR | pre | severe | known | homoz | <i>SLC26A4</i> | c.1198delT | p.(Cys400Valfs*32) | chr7:107330617 |
| FB | Hebron | yes | multi | 3 | AR | pre | yes moderate w/Pendred syn | known | homoz | <i>SLC26A4</i> | c.1198delT | p.(Cys400Valfs*32) | chr7:107330617 |
| J6-8 | Hebron | yes | multi | 2 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.1198delT | p.(Cys400Valfs*32) | chr7:107330617 |
| GR | Nablus | no | multi | 2 | AR | pre | | known | cpd het | <i>SLC26A4</i> | c.[(1198delT);(1001G>T)] | p.[(Cys400Valfs*32);(Gly334Val)] | chr7:107330617; chr7:107323982 |
| CB | Bethlehem | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1341+1delG | p.(?) | chr7:107334926 |
| FU4 | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1341+1delG | p.(?) | chr7:107334926 |
| HB | Hebron | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>SLC26A4</i> | c.1341+1delG | p.(?) | chr7:107334926 |
| X2 | Jersualem | yes | sing | 1 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.1341+1delG | p.(?) | chr7:107334926 |
| a21 | Hebron | yes | sing | 1 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.1341+1delG | p.(?) | chr7:107334926 |
| HF | Nablus | no | multi | 4 | AR | pre | yes severe to profound w/EVA | new | cpd het | <i>SLC26A4</i> | c.[(1341+1delG);(1149_1149+5)] | p.(?);(?) | chr7:107334926; chr7:107329645 |
| Q-B | Tulkarm | yes | multi | 3 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.1489G>A | p.(Gly497Ser) | chr7:107336429 |
| QS002 | Qalqilia | yes | multi | 2 | AR | pre | moderate | known | homoz | <i>SLC26A4</i> | c.2048T>C | p.(Phe683Ser) | chr7:107344789 |
| QS058-059 | Qalqilia | yes | multi | 4 | AR | pre | | known | homoz | <i>SLC26A4</i> | c.2048T>C | p.(Phe683Ser) | chr7:107344789 |
| GA-T | Gaza | yes | multi | 4 | AR | pre | | known | homoz | <i>STRC</i> | complete gene deletion | chr15:43891761_43910998 or larger | |
| CZ9-10 ($\alpha 6-\alpha 7$) | Hebron | yes | multi | 2 | AR | pre | severe to profound | new | homoz | <i>TBC1D24</i> | c.1114_1116del | p.(Ser372del) | chr16:2548369 |
| HD | Hebron | yes | multi | 2 | AR | pre | moderate to severe | known | homoz | <i>TECTA</i> | c.1705C>T | p.(Gln569*) | chr11:120966512 |
| BI | Hebron | yes | multi | 5 | AR | pre | moderate | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| BQ | Bethlehem | yes | multi | 5 | AR | pre | moderate | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| CH | Bethlehem | yes | multi | 7 | AR | pre | moderate to severe | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| CV | Hebron | yes | multi | 11 | AR | pre | moderate | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| EP40 | Bethlehem | yes | multi | 2 | AR | pre | moderate to severe | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| HM | Bethlehem | yes | multi | 4 | AR | pre | moderate to severe | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| HP | Bethlehem | yes | multi | 7 | AR | pre | moderate | known | homoz | <i>TECTA</i> | c.4857C>A | p.(Cys1619*) | chr11:121031011 |
| JA | Hebron | no | multi | 7 | AD | pre | moderate to severe | known | het | <i>TECTA</i> | c.5609A>G | p.(Y470W) | chr11:121038785 |
| GA-P | Gaza | yes | multi | 3 | AR | pre | profound | known | homoz | <i>TMC1</i> | c.100C>T | p.(Arg34*) | chr9:75309494 |
| FA | Nablus | no | multi | 2 | AR | pre | moderate to profound | known | cpd het | <i>TMC1</i> | c.[(100C>T);(1532C>A)] | p.(Arg34*);(Pro511His)] | chr9:75309494; chr9:75407234 |
| GA-F | Gaza | yes | multi | 3 | AR | pre | severe to profound | new | homoz | <i>TMC1</i> | c.1184delA | p.(Gln395Argfs*15) | chr9:75404193 |
| DN | Hebron | yes | multi | 2 | AR | pre | severe to profound | new | homoz | <i>TMC1</i> | c.1763+1G>T | p.(?) | chr9:75431127 |
| AU | Hebron | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| BG-B | Hebron | yes | multi | 9 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| BG-G | Hebron | yes | multi | 7 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| CU | Hebron | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| EP45 | Bethlehem | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| IX | Hebron | yes | multi | 4 | AR | pre | moderate | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| JC | Hebron | yes | multi | 2 | AR | pre | moderate | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| R | Hebron | yes | sing | 1 | AR | pre | moderate to severe | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| W | Hebron | yes | multi | 15 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1193delA | p.(Gln398Argfs*18) | chr21:43796651 |
| CL | Ramallah | yes | multi | 4 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.1176del8ins~1224 | p.(?) | chr21:43796665 |
| EP36 | Bethlehem | yes | sing | 1 | AR | pre | | known | homoz | <i>TMPRSS3</i> | c.989delA | p.(Glu330Glyfs*28) | chr21:43800285 |
| GT | Hebron | yes | sing | 1 | AR | pre | moderate to severe | known | homoz | <i>TMPRSS3</i> | c.989delA | p.(Glu330Glyfs*28) | chr21:43800285 |
| JB | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.989delA | p.(Glu330Glyfs*28) | chr21:43800285 |
| I | Hebron | yes | multi | 3 | AR | pre | | known | homoz | <i>TMPRSS3</i> | c.582T>A | p.(Cys194*) | chr21:43804113 |
| AA-X | Jenin | yes | multi | 3 | AR | pre | severe to profound | known | homoz | <i>TMPRSS3</i> | c.323-6G>A | p.(?) | chr21:43808641 |
| AE | Hebron | yes | sing | 1 | AR | pre | moderate to severe | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| AK | Bethlehem | yes | multi | 6 | AR | pre | moderate to severe | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| AX | Bethlehem | yes | multi | 4 | AR | pre | moderate | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| AZ | Bethlehem | yes | multi | 6 | AR | pre | moderate | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| DU | Hebron | yes | sing | 1 | AR | pre | severe | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| EP30 | Bethlehem | yes | sing | 1 | AR | pre | profound | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| FV | Hebron | yes | multi | 2 | AR | pre | severe | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| ID | Bethlehem | yes | multi | 3 | AR | pre | | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| K | Bethlehem | yes | multi | 11 | AR | pre | severe to profound | known | homoz | <i>TRIOBP</i> | c.1039C>T | p.(Arg347*) | chr22:38119602 |
| HA | Jersualem | yes | multi | 8 | AR | pre | severe to profound | known | cpd het | <i>TRIOBP</i> | c.[(1039C>T);(1741C>T)] | p.[(Arg347*);(Gln581*)] | chr22:38119602; chr22:38120304 |
| X7-X25 | Jersualem | yes | multi | 2 | AR | pre | severe to profound | known | cpd het | <i>TRIOBP</i> | c.[(1039C>T);(1741C>T)] | p.[(Arg347*);(Gln581*)] | chr22:38119602; chr22:38120304 |
| AY | Bethlehem | yes | multi | 9 | AR | pre | moderate | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| BD | Bethlehem | yes | multi | 2 | AR | pre | severe to profound | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| DR | Bethlehem | yes | multi | 4 | AR | pre | profound | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| EP44 | Bethlehem | yes | sing | 1 | AR | pre | moderate to severe | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| FK | Hebron | yes | sing | 1 | AR | pre | severe to profound | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| GE | Hebron | yes | sing | 1 | AR | pre | | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |
| GN | Hebron | yes | multi | 5 | AR | pre | severe | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 |

| Family | City | Consang | Single / multi | | Aff in family | Inherit | Onset pre-post-lingual | | New / known allele | Geno type | Gene | cDNA | Protein | hg19 position | |
|-------------|-----------|---------|----------------|---|---------------|---------|--------------------------------|----------------------------|--------------------|-----------|---------------|------------------|---------------------|-------------------------|----------------|
| | | | HL | | | | Synd | Hearing loss | | | | | | | |
| QS033 | Qalqilia | yes | multi | 3 | AR | pre | severe | | known | homoz | <i>TRIOBP</i> | c.1741C>T | p.(Gln581*) | chr22:38120304 | |
| CM | Hebron | yes | multi | 3 | AR | pre | | | new | homoz | <i>TRIOBP</i> | c.2202_2203delTC | p.(Arg735Glnfs*25) | chr22:38120765 | |
| GM | Jerusalem | yes | multi | 4 | AR | pre | severe to profound | | new | homoz | <i>TRIOBP</i> | c.3232dup | p.(Arg1078Profs*6) | chr22:38121795 | |
| DV | Hebron | yes | multi | 2 | AR | pre | severe to profound, onset 1.5y | | new | homoz | <i>USHIC</i> | c.104+5G>C | p.? | chr11:17554797 | |
| FL | Jerusalem | yes | multi | 2 | AR | pre | yes | profound w/ Usher syndrome | | new | homoz | <i>USHIC</i> | c.104+5G>C | p.? | chr11:17554797 |
| BG-R | Hebron | yes | multi | 3 | AR | pre | yes | profound w/ Usher syndrome | known | homoz | <i>USHIG</i> | c.832_851del | p.(Ser278Profs*71) | chr17:72916080_72916099 | |
| α 14 | Hebron | yes | sing | 1 | AR | pre | | | known | homoz | <i>USHIG</i> | c.469dup | p.(His157Profs*199) | chr17:72916461 | |
| C32 | Hebron | yes | multi | 2 | AR | pre | moderate, no other signs | | new | homoz | <i>USH2A</i> | c.9685delG | p.(Glu3229Argfs*72) | chr1:215987132 | |
| IR | Hebron | yes | multi | 3 | AR | pre | yes | moderate to severe w/RP | | new | homoz | <i>USH2A</i> | c.9685delG | p.(Glu3229Argfs*72) | chr1:215987132 |
| JJ | Hebron | yes | multi | 4 | AR | pre | moderate, no other signs | | new | homoz | <i>USH2A</i> | c.9685delG | p.(Glu3229Argfs*72) | chr1:215987132 | |

Table S3. Hearing loss in consanguineous and non-consanguineous families

| Hearing loss | Consanguineous families | Non consanguineous families | Total |
|---------------------------------|----------------------------|-----------------------------------|-------|
| Autosomal recessive, homozygous | 293 | 2 | 295 |
| AR compound heterozygous | 11 | 17 | 28 |
| Autosomal dominant | 6 | 5 | 11 |
| X-linked | 2 | 1 | 3 |
| No genetic diagnosis | 128 | 26 | 154 |
| Total | 440 | 51 | 491 |

Table S4. Missense mutations in Palestinian families with hearing loss

| Gene | Protein | cDNA | zygosity | hg19 position | Hearing loss phenotype | Pal families with mutation | Pal control exomes (2618) | gnomad exomes (246366) | Our suggestion for ClinVar | Evidence for interpretation in addition to cosegregation P-values and control allele frequencies |
|---|----------------|------------|----------|----------------|---|----------------------------|---------------------------|------------------------|----------------------------|--|
| A. Missense mutations not reported elsewhere: private or Palestinian founder mutations | | | | | | | | | | |
| <i>CACNA1D</i> | p.(Ala376Val) | c.1127C>T | homoz | chr3:53707750 | congenital, moderate w/cardiac signs (prolonged AV conduction on EEG) | 4 | 2.58E-05 | 0 | 4 | LP |
| <i>CDH23</i> | p.(Asp228Val) | c.683A>T | homoz | chr10:73330605 | congenital, profound | 1 | 0.047 | 0 | 0 | LP |
| <i>CDH23</i> | p.(Asn1521Ser) | c.4562A>G | homoz | chr10:73500652 | congenital, profound | 1 | 6.50E-03 | 0 | 5 | LP |
| <i>CDH23</i> | p.(Glu1917Lys) | c.5749G>A | homoz | chr10:73545424 | congenital with RP | 1 | 0.012 | 0 | 0 | LP |
| <i>CDH23</i> | p.(Asp2714Tyr) | c.8140G>T | homoz | chr10:73566000 | congenital, severe to profound | 1 | 0.047 | 0 | 0 | LP |
| <i>CDH23</i> | p.(Leu2735Pro) | c.8204T>C | homoz | chr10:73567059 | congenital, severe to profound | 2 | 1.53E-05 | 0 | 0 | LP |
| <i>COL11A2</i> | p.(Gly1027Glu) | c.3080G>A | homoz | chr6:33139560 | HL and joint problems | 1 | 5.00E-04 | 0 | 0 | LP |
| <i>DNAH11</i> | p.(Thr3824Pro) | c.11470A>C | homoz | chr7:21904249 | severe to profound, recurrent ear infections, conductive? | 1 | 0.026 | 0 | 0 | LP |
| <i>EDNRB</i> | p.(Val275Met) | c.823G>A | homoz | chr13:78477673 | Waardenburg syn | 1 | 0.011 | 0 | 33 | LP |
| <i>ESRRB</i> | p.(Arg182His) | c.545G>A | homoz | chr14:76948389 | congenital, severe to profound | 1 | 6.50E-03 | 1 | 0 | LP |
| <i>LHFPLS</i> | p.(Met1Val) | c.1A>G | homoz | chr6:35773448 | congenital, severe to profound | 1 | 2.44E-04 | 0 | 28 | LP |
| <i>MYO15A</i> | p.(Ala408Val) | c.1223C>T | cpd het | chr17:18023337 | congenital profound | 1 | 0.063 | 9 | 5 | VUS |
| <i>MYO6</i> | p.(Leu926Gln) | c.2777T>A | homoz | chr6:76599892 | congenital, profound | 3 | 3.83E-06 | 0 | 0 | LP |
| <i>MYO7A</i> | p.(Arg895Cys) | c.2683C>T | homoz | chr11:76891516 | congenital, severe to profound | 1 | 6.50E-04 | 0 | 4 | LP |
| <i>OTOA</i> | p.(Asp342Val) | c.1025A>T | homoz | chr16:21716534 | moderate to profound | 2 | 2.50E-08 | 2 | 0 | LP |
| <i>OTOF</i> | p.(Arg1583Cys) | c.1477C>T | homoz | chr2:26688592 | congenital, profound | 1 | 0.036 | 0 | 5 | LP |
| <i>PAX3</i> | p.(Arg68Trp) | c.202C>T | het | chr2:223161816 | Waardenburg syn | 1 | sing | 0 | 0 | LP |
| <i>PCDH15</i> | p.(Asp144Glu) | c.432C>G | homoz | chr10:56128937 | congenital, severe to profound | 1 | 0.047 | 0 | 0 | LP |
| B. Missense mutations suggested to be reclassified from VUS or benign to likely pathogenic | | | | | | | | | | |
| <i>CDH23</i> | p.(Pro2205Leu) | c.6614C>T | homoz | chr10:73553299 | congenital, moderate to severe | 2 | 0.063 | 0 | 0 | LP |
| <i>CLDN14</i> | p.(Pro28Leu) | c.83C>T | homoz | chr21:37833911 | congenital, severe to profound | 7 | 2.50E-10 | 0 | 3 | LP |
| <i>GIPC3</i> | p.(Thr41Lys) | c.122C>A | homoz | chr19:3585717 | severe to profound w/ epilepsy | 1 | 0.02 | 0 | 2 | LP |
| <i>MYO7A</i> | p.(Arg373Cys) | c.1117C>T | homoz | chr11:76871245 | congenital, severe to profound | 1 | 1.72E-05 | 0 | 0 | LP |
| <i>POU3F4</i> | p.(Arg282Leu) | c.845G>T | hemiz | chrX:82764177 | congenital, moderate to severe | 2 | 0.001 | 0 | 0 | LP |
| <i>SLC26A4</i> | p.(Phe354Ser) | c.1061T>C | homoz | chr7:107329557 | congenital, severe to profound | 3 | 6.50E-03 | 5 | 184 | LP |

| Gene | Protein | cDNA | zygosity | hg19 position | Hearing loss phenotype | Pal families with mutation | Pal control exomes (2618) | gnomad exomes (246366) | Our suggestion for ClinVar | Evidence for interpretation in addition to cosegregation P-values and control allele frequencies |
|---|----------------|-----------|----------|-----------------|---|----------------------------|---------------------------|------------------------|----------------------------|--|
| C. Missense mutations previously reported from multiple sources as pathogenic or likely pathogenic | | | | | | | | | | |
| <i>CDH23</i> | p.(Pro346Ser) | c.1036C>T | homoz | chr10:73377052 | congenital, severe to profound | 3 | 0 | 0 | P | |
| <i>CDH23</i> | p.(Pro346Ser) | c.1037C>T | homoz | chr10:73377053 | congenital, moderate | 1 | 0 | 5 | P | |
| <i>CDH23</i> | p.(Pro559Ser) | c.1675C>T | homoz | chr10:73437373 | congenital, moderate to severe | 2 | 0 | 0 | P | |
| <i>CDH23</i> | p.(Glu1071Lys) | c.3211G>A | homoz | chr10:73468959 | congenital, moderate | 2 | 0 | 0 | P | |
| <i>COCH</i> | p.(Cys542Ser) | c.1625G>C | het | chr14:31358969 | onset ~age 20, moderate, progressive | 1 | 0 | 0 | P | |
| <i>ESPN</i> | p.(Ser719Arg) | c.2155A>C | het | chr1:6511986 | | 1 | 0 | 1 | P | |
| <i>GJB2</i> | p.(Trp77Arg) | c.229T>C | homoz | chr13:20763492 | congenital, severe to profound | 1 | 0 | 10 | P | Disulfide bond region, disrupts channel fcn |
| <i>LARS2</i> | p.(Asn153His) | c.457A>C | homoz | chr3:45461162 | severe to profound with ovarian dys and neurological problems | 4 | 1 | 0 | LP | |
| <i>MYO6</i> | p.(Glu299Asp) | c.897G>T | homoz | chr6:76554694 | congenital, profound | 1 | 0 | 0 | P | |
| <i>MYO7A</i> | p.(Gly1298Arg) | c.3892G>A | homoz | chr11:76901883 | | 1 | 2 | 0 | LP | |
| <i>MYO7A</i> | p.(Gly2163Ser) | c.6487G>A | homoz | chr11:76924953 | congenital, moderate to profound | 6 | 0 | 7 | P/LP | |
| <i>MYO15A</i> | p.(Val2114Met) | c.6340G>A | homoz | chr17:18049252 | congenital, severe to profound | 1 | 1 | 1 | P | Also Yang 2013 (compound het with frameshift); no splice effect |
| <i>MYO15A</i> | p.(Arg2728His) | c.8183G>A | homoz | chr17:18058028 | congenital, severe to profound | 3 | 1 | 48 | LP | Completely conserved site; compound het w/fs on ClinVar and in our cohort; no splice effect |
| <i>PAX3</i> | p.(Arg271His) | c.812G>A | het | chr2:223086087 | Waardenburg syn type I | 1 | 0 | 0 | P | |
| <i>PAX3</i> | p.(Ser84Phe) | c.251C>T | het | chr2:223161767 | Waardenburg syn type I | 3 | 0 | 0 | P | |
| <i>PTRH2</i> | p.(Gln86Pro) | c.254T>G | homoz | chr17: 57775086 | severe HL w/mild DD | 2 | 0 | 0 | P/LP | |
| <i>SLC26A4</i> | p.(Ser133Thr) | c.397T>A | homoz | chr7:107312675 | congenital, severe to profound | 1 | 0 | 1 | LP | Missense on ClinVar; we predict enhancer effect |
| <i>SLC26A4</i> | p.(Val239Asp) | c.716T>A | homoz | chr7:107315505 | congenital, severe to profound | 1 | 1 | 51 | P | Damages protein fcn |
| <i>SLC26A4</i> | p.(Gly497Ser) | c.1489G>A | homoz | chr7:107336429 | congenital, severe to profound | 1 | 0 | 7 | P | Damages protein fcn |
| <i>SLC26A4</i> | p.(Phe683Ser) | c.2048T>C | homoz | chr7:107344789 | congenital, moderate | 2 | 0 | 2 | P | |
| <i>TECTA</i> | p.(Tyr1870Cys) | c.5609A>G | het | chr11:121038785 | congenital, moderate to severe | 1 | 0 | 0 | P/LP | Original DFNA8/12 mutation |

Table S5. Mutations with transcript effects. Predictions using approach of Casadei et al 2019. Effects were experimentally validated for genes expressed in available tissues

| Gene | cDNA | Protein | hg19 position | NNSPLICE | | | MaxEnt | | | distance to splice sites | Mutation effect(s) | Transcript effect | Protein effect | Expt confirm in RNA | Families with mutation gnomad | | |
|---------|--------------------|----------------|----------------|----------|--------|---------|--------|--------|---------|--------------------------|--|------------------------------|---|---------------------|-------------------------------|----|----|
| | | | | ref | mutant | cryptic | ref | mutant | cryptic | | | | | | Co-seg | P | |
| ADGRV1 | c.2898G>A | p.(Glu966Glu) | chr5:89940686 | 1.00 | 0.88 | | 8.39 | 3.59 | | EI164I1 | disrupt splice donor exon 15 | del 164 bp | stop codon 931 | yes | 0.002 | 2 | 0 |
| ADGRV1 | c.10426 G>A | p.(Gly3476Arg) | chr5:90024750 | 0.96 | 0.26 | 0.58 | 6.95 | 2.17 | 5.35 | El265I1 | cryptic splice donor c.10216, exon 49 (chr5:90,024,540) | del 210 bp | del aa 3407-3476 in EAR4 domain | yes | 0.012 | 1 | 0 |
| CDC14A | c.51-1G>T | p.(?) | chr1:100819317 | 0.87 | 0.00 | | 11.01 | 2.41 | | | disrupt splice acceptor exon 2 | del 91 bp | stop codon 25 | yes | sing | 1 | 0 |
| CLPP | c.270+3G>T | p.(?) | chr19:6361954 | 0.96 | 0.54 | | 8.69 | 1.83 | | | disrupt splice donor exon 2; retain intron 2 | del 72 bp; ins 505 bp | del aa 67-90 Clp protease proteolytic subunit; stop cdn 128 | na | 0.105 | 1 | 0 |
| DFNA5 | c.991_15_991-13del | p.(?) | chr7:24746008 | | | | | | - | | weaken polypyrimidine tract at exon 8 acceptor | del 193 bp | stop codon 372 | yes | to do | 1 | |
| EDNRB | c.1071+2T>C | p.(?) | chr13:78477289 | 0.90 | 0.00 | | 8.76 | 1.01 | | | disrupt splice donor exon 4 | del 205 bp | stop codon 317 | yes | sing | 1 | 0 |
| ESRRB | c.787G>A | p.(Gly263Ser) | chr14:76949102 | 1.00 | 0.91 | | 9.60 | 6.39 | | El162I1 | disrupt splice donor exon 7 | del 162 bp | del aa 209-262 in transactivation domain | yes | 0.001 | 2 | 1 |
| GJB2 | c.-23+1G>A | p.(?) | chr13:20766921 | 0.99 | 0.00 | | 9.60 | 1.42 | | | disrupt splice donor exon 1 | loss of transcript | no protein | yes | | 4 | |
| HSD17B4 | c.1842G>A | p.(Lys614Lys) | chr5:118862914 | 0.99 | 0.56 | 0.95 | 9.11 | 3.87 | 4.82 | El87I1 | disrupt splice donor exon 21; cryptic splice donor c.1842+70 | del 87 bp | del aa 586-614 in hydratase domain; stop codon 616 | yes | 0.141 | 1 | 5 |
| LHFPL5 | c.*16+1G>A | p.(?) | chr6:35787241 | 0.98 | 0.00 | | 11.11 | 2.93 | | | disrupt splice donor exon 3 (3'UTR) | del 12 bp | extend 32 codons (PMID:30177809) | known allele | sing | 1 | 0 |
| LRTOMT | c.327C>T | p.(Cys109Cys) | chr11:71817225 | 0.94 | | 0.83 | 8.68 | | 7.39 | El244I32 | cryptic splice donor at c.326 in exon 7 | del 33 bp | del aa 110-120 in transmembrane domain | ne | 0.011 | 1 | 0 |
| MYO6 | c.897G>T | p.(Glu299Asp) | chr6:76554694 | 0.36 | 0.00 | | 7.13 | -2.49 | | El81I1 | disrupt splice donor exon 10 | del 81 bp | del aa 275-301 in ATPase domain | yes | 9.77E-04 | 2 | 0 |
| MYO7A | c.4951G>A | p.(Gly1651Ser) | chr11:72912591 | | | | | | | El99I93 | ESE loss and ESS gain exon 36 | del 191 bp | stop codon 1656 | yes | 0.079 | 1 | 0 |
| MYO7A | c.4153-2A>G | p.(?) | chr11:76905397 | 0.88 | 0.00 | | 5.93 | -2.02 | | | disrupt splice acceptor exon 32 | del 171 bp | del aa 1385-1441 in FERM1 domain | known allele | | 3 | |
| MYO15A | c.3609+985G>A | p.(?) | chr17:18026708 | 0.96 | 0.00 | | 7.77 | -0.41 | | | disrupt splice donor exon 1 | comp loss | cochlear and pituitary-specific isoform | ne | | 9 | 0 |
| MYO15A | c.3866+1G>A | p.(?) | chr17:18029771 | 0.93 | 0.00 | | 8.40 | 0.22 | | | disrupt splice donor exon 5 | 110 bp | stop codon 1277 | ne | sing | 1 | 2 |
| MYO15A | c.4142+5G>A | p.(?) | chr17:18034661 | 0.97 | 0.09 | 0.54 | 9.25 | 2.65 | 6.42 | | disrupt splice donor exon 9; cryptic donor splice exon 9 | 106 bp; del 70 bp | stop codon 1360 | ne | 0.016 | 2 | 0 |
| MYO15A | c.4240G>A | p.(Glu1414Lys) | chr17:18035800 | | | | | | | El34I81 | ESS gain exon 11 | del 114 bp | del aa 1403-1440 in motor domain | ne | 7.32E-04 | 3 | 1 |
| MYO15A | c.6499T>C | p.(Tyr2167His) | chr17:18049411 | | | | | | | El226I11 | ESE loss exon 30 | del 236 bp | stop codon 2095 | ne | sing | 1 | 7 |
| MYO15A | c.6893G>A | p.(Arg2298Gln) | chr17:18052203 | | | | | | | El129I64 | ESE loss, ESS gain, exon 33 | del 192 bp | del aa 2256-2319 in tail domain | ne | 0.002 | 1 | 2 |
| MYO15A | c.7207G>T | p.(Asp2403Tyr) | chr17:18052889 | 0.00 | 0.87 | | 0.08 | 7.73 | | El90I6 | cryptic splice donor c.7206, exon 35 (chr17:18,052,888) | del 7 bp | stop codon 2414 | ne | 4.67E-17 | 13 | 0 |
| MYO15A | c.8309_8311del | p.(Glu2770del) | chr17:18058506 | | | | | | | El84I31 | ESE loss exon 46 | del 116 bp | stop codon 2803 | ne | 0.026 | 1 | 0 |
| MYO15A | c.8788+5G>T | p.(?) | chr17:18060549 | 0.38 | 0.00 | | 3.29 | -3.85 | | | disrupt splice donor exon 50 | del 75 bp | del aa 2906-2930 in SH3 domain | ne | 0.105 | 1 | 0 |
| MYO15A | c.9572G>A | p.(Arg3191His) | chr17:18065953 | | | | | | | El55I41 | ESE loss, ESS gain, exon 58 | del 95 bp | stop codon 3189 | ne | 0.008 | 2 | 10 |
| OTOA | c.2017-2A>G | p.(?) | chr16:21739560 | 0.71 | 0.00 | 0.30 | 7.47 | -0.48 | 5.22 | | disrupt splice acceptor exon 20; cryptic splice acceptor c.2017-72 | del 191 bp; ins 71 bp | stop codon 696; stop codon 751 | yes | 0.016 | 2 | 0 |
| SLC26A4 | c.284G>A | p.(Gly95Glu) | chr7:107303860 | | | | | | | El120I21 | ESE loss; ESS gain exon 3 | del 140 bp | stop codon 134 | yes | 0.105 | 1 | 0 |
| SLC26A4 | c.304G>A | p.(Gly102Arg) | chr7:107303880 | 0.99 | 0.82 | | 10.57 | 6.91 | | El140I1 | disrupt splice donor exon 3 | del 140 bp | stop codon 134 | yes | 0.002 | 1 | 0 |
| SLC26A4 | c.397T>A | p.(Ser133Thr) | chr7:107312675 | | | | | | | El93I19 | ESE loss exon 4 | del 111 bp | del aa 102-138 in TM domain | yes | 0.016 | 1 | 0 |
| SLC26A4 | c.1001G>T | p.(Gly334Val) | chr7:107323982 | 0.98 | 0.45 | 0.94 | 7.93 | 6.05 | 9.14 | El83I1 | (1) disrupt splice donor exon 8; (2) cryptic splice donor | (1) del 83 bp; (2) ins 40 bp | (1) stop codon 311; (2) stop codon 355 | yes | 6.86E-11 | 10 | 2 |
| SLC26A4 | c.1341+1delG | p.(?) | chr7:107334926 | | | | 7.08 | -5.81 | | El1178 | disrupt splice donor exon 11 | del 78 bp | del aa 422-446, complete TM domain | yes | | 5 | 4 |
| TBC1D24 | c.1114_1116del | p.(Ser372del) | chr16:2548369 | | | | | | | El131I27 | ESE loss exon 4 | del 159 bp | del aa 328-380 in TBC domain | yes | 0.047 | 1 | 0 |
| TMCI | c.1763+1G>T | p.(?) | chr9:75431127 | 0.52 | 0.00 | | 6.99 | -1.51 | | | disrupt splice donor exon 19 | del 68 bp | stop codon 600 | known allele | sing | 1 | 0 |
| USH1C | c.104+5G>C | p.(?) | chr11:17554797 | 0.99 | 0.23 | | 9.09 | 1.17 | | | disrupt splice donor exon 2 | del 68 bp | stop codon 29 | ne | 0.004 | 2 | 0 |

ne: not expressed in available tissue

na: family not available

Table S6. Unpaired heterozygous mutations in probands with apparently recessive hearing loss

| Gene | cDNA | Chr | position (hg19) | Type | Family | Family type | Consang | Gene solving family |
|---|------------------|---------------------|--------------------|----------------------|--------|----------------|---------|------------------------|
| A. Families with no genetic diagnosis | | | | | | | | |
| <i>GJB2</i> | p.(Thr8Met) | c.23C>T (exon2) | 13 | 20,763,698 missense | JM | sing | yes | not solved |
| <i>GJB2</i> | p.(Gly12Valfs*2) | c.35delG (exon2) | 13 | 20,763,686 delFS | A158 | sing | no | not solved |
| <i>GJB2</i> | p.(Gly12Valfs*2) | c.35delG (exon2) | 13 | 20,763,686 delFS | AS-HH | sing | no | not solved |
| <i>GJB2</i> | p.(Gly12Val) | c.35G>T (exon2) | 13 | 20,763,686 missense | AS-II | sing | no | not solved |
| <i>GJB2</i> | p.(Tyr65Ser) | c.194A>C (exon2) | 13 | 20,763,527 missense | HZ | sing | yes | not solved |
| <i>GJB2</i> | p.(Arg98Trp) | c.292C>T (exon2) | 13 | 20,763,429 missense | HW | sing | yes | not solved |
| <i>GJB2</i> | p.(Met163Val) | c.487A>G (exon2) | 13 | 20,763,234 missense | H | sing | yes | not solved |
| <i>MYO15A</i> | p.(Arg2728His) | c.8183G>A (exon44) | 17 | 18,058,028 missense | GD | sing | no | not solved |
| <i>OTOG</i> | p.(Arg2485fs*) | c.7454delG (exon43) | 11 | 17,655,766 delFS | A173 | sing | yes | not solved |
| <i>SLC26A5</i> | - | c.971+2T>C (exon9) | 7 | 103,038,377 splice | A173 | sing | yes | not solved |
| <i>TECTA</i> | p.(Cys1619*) | c.4857C>A (exon14) | 11 | 121,031,011 nonsense | FC14 | multi | yes | not solved |
| <i>USH2A</i> | p.(Gly2313Cys) | c.6937G>T (exon36) | 1 | 216,143,987 missense | GA-M | sing | yes | not solved |
| B. Families with hearing loss solved by a different gene | | | | | | | | |
| <i>GJB2</i> | | c.-23+1A>C | 13 | 20,763,744 splice | IW | multi | yes | <i>EPS8L2</i> |
| <i>GJB2</i> | p.(Gly12Valfs*2) | c.35delG (exon2) | 13 | 20,763,686 delFS | AS-T | multi | yes | <i>PCDH15</i> |
| <i>GJB2</i> | p.(Gly12Valfs*2) | c.35delG (exon2) | 13 | 20,763,686 delFS | QS059 | multi | yes | <i>SLC26A4</i> |
| <i>GJB2</i> | p.(Gly12Val) | c.35G>T (exon2) | 13 | 20,763,686 missense | JF | multi | yes | <i>SLC26A4</i> |
| <i>GJB2</i> | p.(Leu79Cysfs*3) | c.235delC (exon2) | 13 | 20,763,486 delFS | GR | multi | yes | <i>SLC26A4</i> |
| <i>GJB2</i> | p.(Leu90Pro) | c.269T>C (exon2) | 13 | 20,763,452 missense | AA-N | multi | yes | <i>CDH23</i> |
| <i>GJB2</i> | p.(Tyr97*) | c.290dupA (exon2) | 13 | 20,763,430 nonsense | AS-R | sing | yes | <i>MYO7A</i> |
| <i>GJB2</i> | p.(Met163Val) | c.487A>G (exon2) | 13 | 20,763,234 missense | JH | multi | no | <i>MYO7A</i> |
| <i>GJB2</i> | p.(Met163Val) | c.487A>G (exon2) | 13 | 20,763,234 missense | QS043 | sing | yes | <i>MYO7A</i> |