### Additional file 9: Variants of novel candidate genes associated with hearing loss.

| Family ID | Gene symbol | Tier | Reference mRNA | Nucleotide change (GRCh37) | Amino acid change | Genomic position (GRCh37) | dbSNP ID | MAF (1000G) | MAF (ESP6500) | MAF (ExAC) | MAF (gnomAD) | MAF (HGVD) | MAF (in house) | REVEL | CADD | UCSF conservation | Splice site (MaxEntScan) | Splice site (HSF3.0) | Splice site Affected |
|-----------|-------------|------|----------------|-----------------------------|------------------|---------------------------|-----------|--------------|--------------|-------------|-------------|-------------|----------------|---------|-------|------------------|---------------------|---------------------|------------------|
| 1427      | BAIAP2L2    | 2    | NM_025045.5    | c.506T>C                    | p.(Val169Ala)    | chr22:38493145A>G        | none      | 0            | 0            | 0           | 0           | 0           | 0.287          | 25.8    | 11/11 | 42/48, 20/27    | -                   | -                   | -31.14%          |
| 1676      | HKDC1       | 3    | NM_025130.3    | c.376-2A>G                  | p.? (splice mut) | chr10:70992768A>G        | none      | 0            | 0            | 0           | 0           | 0           | -             | 33.0    | -     | -                | 11.49 to 3.54       | -                   | -                |
| 1676      | HKDC1       | 3    | NM_025130.3    | c.1771A>C                   | p.(Lys591Gln)    | chr10:71010343A>C        | rs376814835 | 0            | 8.242E-06    | 0           | 0           | 0           | 0.708          | 23.2    | 12/12 | 47/48, 36/37    | -                   | -                   | -                |
| 1535      | SVEP1       | 4    | NM_153366.3    | c.6766C>G                   | p.(Pro2256Ala)   | chr9:113717114G>C        | rs369710771 | none         | 0            | 0           | 0           | 0           | 0.589          | 25.0    | 11/11 | 50/50, 35/36    | -                   | -                   | -                |
| 1535      | SVEP1       | 4    | NM_153366.3    | c.7357G>A                   | p.(Val2453Met)   | chr9:11370523C>T         | rs374113606 | 0            | 0.000E-04    | 8.273E-05   | 4.819E-05   | 0.002        | 0.264          | 25.6    | 11/11 | 50/50, 37/37    | -                   | -                   | -                |
| 1555      | SVEP1       | 4    | NM_153366.3    | c.4977C>T                   | p.(Pro2328Leu)   | chr9:11370903G>A         | rs369710771 | 0            | 8.300E-05    | 1.160E-04   | 9.274E-05   | 0.001        | 0.589          | 25.0    | 11/11 | 50/50, 35/36    | -                   | -                   | -                |
| 1655      | SVEP1       | 4    | NM_153366.3    | c.10294T>C                  | p.(Tyr3432His)   | chr9:11341741A>G         | none      | 0            | 0            | 0           | 4.018E-06   | 4.132E-04   | 0.235          | 23.0    | 11/11 | 50/50, 20/33    | -                   | -                   | -                |
| 1669      | CACNG1      | 4    | NM_000727.3    | c.461C>T                    | p.(Ser154Leu)    | chr17:65052179C>T        | rs866479653 | none         | 0            | 0           | 0           | 0           | 0.683          | 26.3    | 12/12 | 43/45, 37/37    | -                   | -                   | -                |
| 1696      | GTPBP4      | 4    | NM_012341.2    | c.967C>G                    | p.(Leu323Val)    | chr10:1051812C>G         | none      | 0            | 0            | 0           | 0           | 0           | 0.23           | 16.4    | 12/12 | 48/48, 32/32    | -                   | -                   | -                |
| 1685      | PCNX2       | 4    | NM_014801.3    | c.3506C>T                   | p.(Arg1169Trp)   | chr1:232096041G>A        | rs150235720 | 0            | 0.002        | 0.001       | 1.385E-03   | 0           | 0.139          | 25.5    | 12/12 | 45/47, 20/36    | -                   | -                   | -                |
| 1685      | PCNX2       | 4    | NM_014801.3    | c.4777C>T                   | p.(Arg1593Ter)   | chr1:232157229G>A        | rs751554683 | 0            | 0            | 1.854E-05   | 1.204E-05   | 0           | 0           | 0.625          | 26.7    | -     | 42.0             | -                   | -                   | -                |
| 1575      | TBC1D8      | 4    | NM_001102426.1 | c.1997C>T                   | p.(Ser666Leu)    | chr2:101646133G>A        | none      | 0            | 0            | 4.130E-05   | 4.012E-05   | 0           | 0.569          | 29.4    | 12/12 | 49/49, 36/38    | -                   | -                   | -                |