**Panel-based NGS reveals disease causing mutations in hearing loss patients using BGISEQ-500 platform**

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**Supplementary material**

**Gene list**

GJB2, GJB6, MYO7A, MYO15A, FOXI1, KCNJ10, SLC26A4, TMIE, TMC1, TMPRSS3, OTOF, CDH23, ATP2B2, GIPC3, STRC, OTOG, USH1C, TECTA, OTOA, PCDH15, RDX, GRXCR1, TRIOBP, CLDN14, MYO3A, WHRN, ESRRB, ESPN, MYO6, GJA1, HGF, ILDR1, MARVELD2, DFNB59, SLC26A5, LRTOMT, LHFPL5, BSND, MSRB3, LOXHD1, TPRN, GPSM2, PTPRQ, SERPINB6, GJB3

PRPS1, POU3F4, SMPX

ACTG1, CCDC50, CEACAM16, COCH, CRYM, DFNA5, DIABLO, DIAPH1, DSPP, EYA4, GJB2, GJB3, GJB6, GRHL2, KCNQ4, MIR96, MYH14, MYH9, MYO1A, MYO6, MYO7A, POU4F3, SIX1, SLC17A8, TECTA, TJP2, TMC1, WFS1, DIAPH3

MT-RNR1, MT-TS1

SERAC1, PDSS1, FGFR3, FGFR1, FGFR2, PHEX, DLX5, TNFRSF11B, COL2A1, COL11A1, COL9A1, COL9A2, COL4A3, COL4A4, COL4A5, BSND, SOX9, PAX2, GATA3, SLC19A2, IGF1, PAX3, MITF, SNAI2, EDNRB, EDN3, SOX10, HOXA1, SOBP, EYA1, SIX5, SIX1, CHD7, SEMA3E, SMAD4, FGF3, TCOF1, PRRX1, GLI3, HOXA2, KCNQ1, KCNE1, CACNA1D, ALMS1, LRP2, TIMM8A, NDP, WFS1, OPA1, SLC4A11, MYO7A, USH1C, CDH23, PCDH15, USH1G, USH2A, ADGRV1, PDZD7, WHRN, CLRN1, MT-TK, MT-TE, MT-TL1, SLC26A4, KCNJ10, FOXI1

**Access number for the novel mutations:** all novel mutations identified in this study have been submitted to CNGB Nucleotide Sequence Archive (<https://db.cngb.org/cnsa>) with the following accession numbers:

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| --- | --- | --- | --- | --- |
| **Number** | **Sample Name** | **Mutation** | **Mutation**  **Status** | **Accession number** |
| 1 | HL1 | OTOG (NM\_001277269): c.1190G>A (p.Arg397Gln) | Novel | var030008 |
| 2 | HL4 | SLC26A4 (NM\_000441): c.1519delT (p.Leu507Ter fsX5) | Novel | var030009 |
| 3 | HL5 | SLC26A4 (NM\_000441): c.249G>A (p.Trp83Ter) | Novel | var030010 |
| 4 | HL6 | MYO15A (NM\_016239): c.5134-10C>G | Novel | var030011 |
| 5 | HL8 | SLC26A4 (NM\_000441): c.1519delT (p.Leu507TerfsX5) | Novel | var030009 |
| 6 | HL9 | PTPRQ (NM\_001145026): c.5942+1G>A;  PTPRQ (NM\_001145026): c.6024G>A (p.Ser2008Ser) | Novel;  Novel | var030012  var030013 |
| 7 |
| 8 | HL10 | MYO15A (NM\_016239): c.10420A>G (p.Ser3474Gly) | Novel | var030014 |
| 9 | HL11 | ESPN (NM\_031475): c.1464+2T>A | Novel | var030015 |
| 10 | HL13 | MYO6 (NM\_004999): c.118-2A>G | Novel | var030016 |
| 11 | HL17 | TRIOBP (NM\_001039141): c.2321delG (p.Arg774HisfsX105) | Novel | var030017 |
| 12  13 | HL18 | DIAPH3 (NM\_001042517): c.3431delC (p.Thr1144SerfsX17);  MYO15A (NM\_016239): c.10245\_10247delCTC | Novel;  Novel | var030018  var030019 |
| 14 | HL20 | MT-RNR1 (NC\_012920): m.1119T>C | Novel | var030020 |
| 15 | HL21 | TRIOBP (NM\_001039141): c.3256C>A (p.Pro1086Thr) | Novel | var030021 |
| 16 | HL22 | DSPP (NM\_014208): c.1057delA (p.Lys353AsnfsX3) | Novel | var030022 |
| 17 | HL23 | DIAPH1 (NM\_001079812): c.1982C>T (p.Pro661Leu) | Novel | var030023 |
| 18 | HL24 | MITF (NM\_198159): c.1025\_1032delGGAACAAG (p.Trp342TrpfsX25) | Novel | var030024 |
| 19 | HL25 | SLC26A4 (NM\_000441): c.1339delA (p.Lys447SerfsX8); | Novel; | var030025 |
| 20  21 | HL26 | CDH23 (NM\_022124): c.9640C>A (p.Leu3214Met);  PCDH15 (NM\_033056): c.3807G>T (p.Glu1269Asp) | Novel;  Novel | var030026  var030027 |
| 22 | HL28 | MYO7A (NM\_000260): c.562C>G (p.Gln188Glu) | Novel | var030028 |
| 23 | HL31 | MYO7A (NM\_000260): c.1004C>G (p.Ala335Gly) | Novel | var030029 |
| 24  25 | HL32 | MYO7A (NM\_000260): c.586C>G (p.Leu196Val);  MYO7A (NM\_000260): c.1679A>G (p.Tyr560Cys) | Novel;  Novel | var030030  var030031 |
| 26 | HL33 | KCNQ4 (NM\_172163): c.1905G>A (p.Ser635Ser) | Novel | var030032 |
| 27 | HL36 | EDN3 (NM\_000114): c.142G>A (p.Glu48Lys) | Novel | var030033 |
| 28 | HL37 | MYO15A (NM\_016239 ): c.10245\_10247delCTC | Novel | var030034 |
| 29 | HL38 | TRIOBP (NM\_001039141): c.4429\_4430insG (p.Trp1477TrpfsX25); | Novel | var030035 |
| 30  31  32 | HL40 | KCNQ1 (NM\_000218): c.2016\_2017insGATGAGGGGTCCT (p.Pro672ProfsX6);  TRIOBP (NM\_001039141): c.3943\_3944insCTCTTCGG (p.Arg1315ProfsX44);  TRIOBP (NM\_001039141): c.3941A>C (p.Glu1314Ala) | Novel;  Novel;  Novel | var030036  var030037  var030038 |
| 33 | HL41 | MYO15A (NM\_016239): c.8681\_8682insA (p.Ile2894IlefsX32) | Novel | var030039 |
| 34  35 | HL43 | TRIOBP (NM\_001039141): c.5185-2A>G;  PTPRQ (NM\_001145026): c.3194delT (p.Ile1065IlefsX6) | Novel;  Novel | var030040  var030041 |
| 36 | HL44 | TMC1 (NM\_138691): c.2050G>C (p.Asp684His) | Novel | var030042 |