**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:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **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 | var030012var030013 |
| 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 |
| 1213 | HL18 | DIAPH3 (NM\_001042517): c.3431delC (p.Thr1144SerfsX17); MYO15A (NM\_016239): c.10245\_10247delCTC | Novel;Novel | var030018var030019 |
| 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 |
| 2021 | HL26 | CDH23 (NM\_022124): c.9640C>A (p.Leu3214Met);PCDH15 (NM\_033056): c.3807G>T (p.Glu1269Asp) | Novel;Novel | var030026var030027 |
| 22 | HL28 | MYO7A (NM\_000260): c.562C>G (p.Gln188Glu) | Novel | var030028 |
| 23 | HL31 | MYO7A (NM\_000260): c.1004C>G (p.Ala335Gly) | Novel | var030029 |
| 2425 | HL32 | MYO7A (NM\_000260): c.586C>G (p.Leu196Val);MYO7A (NM\_000260): c.1679A>G (p.Tyr560Cys) | Novel;Novel | var030030var030031 |
| 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 |
| 303132 | 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 | var030036var030037var030038 |
| 33 | HL41 | MYO15A (NM\_016239): c.8681\_8682insA (p.Ile2894IlefsX32) | Novel | var030039 |
| 3435 | HL43 | TRIOBP (NM\_001039141): c.5185-2A>G; PTPRQ (NM\_001145026): c.3194delT (p.Ile1065IlefsX6) | Novel;Novel | var030040var030041 |
| 36 | HL44 | TMC1 (NM\_138691): c.2050G>C (p.Asp684His) | Novel | var030042 |