**Table 2.** Clinical data and genetics evaluations of the diagnosed patients

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Family** | **Patient** | **Sex** | **Age** | **Panel** | **Diagnosis** | **Gene** | **Allele 1** | **Allele 2** | **Onset** | **Severity** | **Family history** |
| **(A) Patients Diagnosed with Autosomal Recessive Deafness** | | | | | | | | | | | |
| 1 | 4377 | F | 9y | HL | NSHL/EVA | *SLC26A4*  (NM\_000441.2) | c.563T>C/(p.Ile188Thr) | c.1547dupC/(p.Ser517PhefsTer10) | Prelingual | Severe | No |
|  | c.1746delG/(p.Ala584ArgfsTer2) |  |
| 2 | 7836 | M | 8y | HL | NSHL/EVA | SLC26A4  (NM\_000441.2) | c.919-2A>G | c.1975G>C/(p.Val659Leu) | Prelingual | Profound | No |
| 3 | 16175 | M | 5y | HL | NSHL/EVA | SLC26A4  (NM\_000441.2) | c.589G>C/(p.Gly197Arg) | c.754T>C/(p.Ser252Pro) | Post-lingual | Severe | No |
| 4 | 50765 | F | 2y | HL | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.235delC/(p.Leu79CysfsTer3) | Prelingual | Profound | Yes |
| 5 | 58589 | M | 26y | HL | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.508\_511dupAACG/(p.Ala171GlufsTer40) | Prelingual | Profound | No |
| 6 | 58592 | F | 11y | HL | NSHL | MYO15A  (NM\_016239.4) | c.3524dupA/(p.Ser1176ValfsTer14) | c.3866+5G>A | Prelingual | Severe | No |
| 7 | 58207 | F | 1y | HL | NSHL | GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) | c.109G>A/(p.Val37Ile) | Prelingual | Moderate | No |
| 8 | 45935 | F | 6y | HL | NSHL | MYO15A  (NM\_016239.4) | c.1203C>A/(p.Tyr401Ter) | c.5649+1G>T | Post-lingual | Moderate | No |
| 9 | 46181 | F | 8y | HL | NSHL | GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) | c.109G>A/(p.Val37Ile) | Prelingual | Moderate | No |
| 10 | 61919 | M | 4y | HL | NSHL | LRTOMT  (NM\_001145308.5) | c.566delT/(p.Ile189ThrfsTer8) | c.655C>T/(p.Arg219Ter) | Prelingual | Profound | No |
| 11 | 68057 | M | 8y | HL | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.235delC/(p.Leu79CysfsTer3) | Prelingual | Severe | No |
| 12 | 57197 | F | 27y | HL | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.139G>T/(p.Glu47Ter) | Prelingual | Severe | Yes |
| 13 | 34030 | F | 3y | CE | USH | CDH23  (NM\_022124.6) | c.1606C>T/(p.Arg536Trp) | c.8371delC/(p.Leu2791TrpfsTer46) | Prelingual | Severe | No |
| 14 | 23697 | M | 42y | CE | NSHL | GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) | c.109G>A/(p.Val37Ile) | Post-lingual | Moderate | No |
| STRC  (NM\_153700.2) | Exon16 deletion |  |
| 15 | 33273 | M | 1y | CE | NSHL | LRTOMT  (NM\_001145308.5) | c.566delT/(p.Ile189ThrfsTer8) | c.566delT/(p.Ile189ThrfsTer8) | Prelingual | Severe | No |
| 16 | 30140 | M | 5y | CE | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.427C>T/(p.Arg143Trp) | Prelingual | Severe | No |
| 17 | 39130 | M | 25y | CE | NSHL/EVA | SLC26A4  (NM\_000441.2) | c.919-2A>G | c.589G>A/(p.Gly197Arg) | Prelingual | Severe | No |
| PMP22  (NM\_000304.3) | Whole gene deletion (17p12) |  |
| 18 | 39131 | F | 48y | CE | NSHL | GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) | c.379C>T/(p.Arg127Cys) | Prelingual | Severe | No |
| 19 | 21979 | M | 27y | CE | NSHL/EVA | SLC26A4  (NM\_000441.2) | c.919-2A>G | c.2086C>T/(p.Gln696Ter) | Prelingual | Profound | No |
| 20 | 21980 | F | 24y | CE | NSHL | GJB2  (NM\_004004.6) | c.235delC/(p.Leu79CysfsTer3) | c.235delC/(p.Leu79CysfsTer3) | Prelingual | Profound | No |
| PTPRQ  (NM\_001145026.2) | c.5217delT/(p.Val1740LeufsTer4) |  |
| 21 | 25248 | M | 11y | CE | NSHL | STRC  (NM\_153700.2) | c.2303\_2313+1del | Exon16-19 duplication | Post-lingual | Moderate | No |
| 22 | 9989 | M | 3y | CE | NSHL | MYO7A  (NM\_000260.4) | c.4398G>A/(p.Trp1466Ter) | c.4485G>A/(p.Trp1495Ter) | Prelingual | Severe | No |
| 23 | 61911 | M | 9y | CE | NSHL | MYO15A  (NM\_016239.4) | c.5275A>C/(p.Ser1759Arg) | c.10250\_10252delCCT/(p.Ser3417del) | Prelingual | Severe | No |
| 24 | 25257 | M | 43y | CE | NSHL | TMC1  (NM\_138691.3) | c.453+2T>C | c.453+2T>C | Prelingual | Profound | No |
| GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) |  |
| 25 | 62102 | F | 34y | HL | NSHL | MYO15A  (NM\_016239.4) | c.3524dupA/(p.Ser1176ValfsTer14) | c.3524dupA/(p.Ser1176ValfsTer14) | Prelingual | Profound | No |
| 26 | 21603 | F | 2y | HL | NSHL | LHFPL5  (NM\_182548.4) | c.619delC/(p.Leu207SerfsTer37) | c.619delC/(p.Leu207SerfsTer37) | Prelingual | Profound | No |
| 27 | 31403 | F | 5y | HL | NSHL | MYO15A  (NM\_016239.4) | c.10250\_10252delCCT/(p.Ser3417del) | c.8138T>G/(p.Leu2713Arg) | Prelingual | Severe | No |
| 28 | 65920 | F | 35y | CE | NSHL | MYO7A  (NM\_000260.4) | c.291T>G/(p.Tyr97Ter) | c.285+5G>C | Post-lingual | Moderate | No |
| GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) |  |
| **(B) Patients Diagnosed with Autosomal Dominant Deafness** | | | | | | | | | | | |
| 29 | 2496 | F | 31y | HL | NSHL | TMC1  (NM\_138691.3) | c.1714G>A/(p.Asp572Asn) |  | Post-lingual | Severe | Yes |
| 29 | 2497 | M | 56y | HL | NSHL | TMC1  (NM\_138691.3) | c.1714G>A/(p.Asp572Asn) |  | Post-lingual | Severe | Yes |
| 30 | 49145 | F | 8y | HL | NSHL | GJB6  (NM\_001110219.3) | c.228delG/(p.Trp77GlyfsTer5) |  | Prelingual | Profound | No |
| 31 | 14216 | M | 7m | CE | Waardenburg syndrome | SOX10  (NM\_006941.4) | c.549\_567del/(p.Gln185ProfsTer95) |  | Prelingual | Severe | No |
| 32 | 51456 | F | 5y | CE | CHARGE syndrome | CHD7  (NM\_017780.4) | c.4353G>C/(p.Gly1451=) |  | Prelingual | Severe | No |
| GJB2  (NM\_004004.6) | c.109G>A/(p.Val37Ile) | c.109G>A/(p.Val37Ile) |
| 33 | 56557 | F | 2y | CE | LEOPARD syndrome | PTPN11  (NM\_002834.5) | c.1403C>T/(p.Thr468Met) |  | Prelingual | Moderate | No |
| 34 | 54210 | M | 1y | HL | NSHL | EYA1  (NM\_000503.6) | c.922C>T/(p.Arg308Ter) |  | Prelingual | Severe | Yes |
| 34 | 54211 | M | 25y | HL | NSHL | EYA1  (NM\_000503.6) | c.922C>T/(p.Arg308Ter) |  | Prelingual | Profound | Yes |
| 35 | 30503 | M | 5y | HL | NSHL | EYA1  (NM\_000503.6) | c.473delC/(p.Pro158LeufsTer83) |  | Prelingual | Severe | No |
| 36 | 39101 | M | 25y | HL | NSHL | MYH14  (NM\_001077186.2) | c.289\_290insTGG/(p.Gln97delinsLeuGlu) |  | Post-lingual | Severe | Yes |
| 36 | 39100 | M | 2y | HL | NSHL | MYH14  (NM\_001077186.2) | c.289\_290insTGG/(p.Gln97delinsLeuGlu) |  | Post-lingual | Mild | Yes |
| 37 | 5428 | F | 44y | HL | NSHL | GSDME  (NM\_001127453.2) | c.991-2A>G |  | Post-lingual | Severe | Yes |