**Table S6**. **GJB2 direct sequencing** – Brief description of patient's phenotype and variants in GJB2 detected by direct sequencing. All patients displayed congenital, SN, bilateral HL. None of them were born from a consanguinous union. F= female, M=male, NR= not reported, rs= Reference SNP.

| Patient | Sex | Severity | Family history | Malformations at the CT/IRM | Additional signs | Variant description | Rs number |
|---|---|---|---|---|---|---|---|
| *71* | M | Severe | No | Yes | NR | homozygous c.35delG, p.(Gly12Valfs*2) in *GJB2* | rs80338939 |
| *72* | M | Severe | No | Not performed | NR | homozygous c.35delG, p.(Gly12Valfs*2) in *GJB2* | rs80338939 |
| *73* | F | Moderate | No | Not performed | NR | compound heterozygous for c.59T>C, p.(Ile20Thr), heterozygous for c.109G>A, p.(Val37Ile) in *GJB2* | rs1057517519; rs146378222 |
| *74* | M | Severe | No | Yes | No | homozygous c.269T>C, p.Leu90Pro in *GJB2* | rs80338945 |
| *75* | F | Profound | Yes | Not performed | No | homozygous c.35delG, p.(Gly12Valfs*2) in *GJB2* | rs80338939 |