

# **Recurrent and Novel Pathogenic Variants in Genes Involved with Hearing loss in the Pakistani Population**

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**Supplementary Table 1. Rare variants identified in the proband of PKHL-21**

| Gene           | Transcript  | cDNA (c.)        | Protein Change (p.) | Zygosity | MAF gnomAD | Hom. gnomAD | MutationTaster | PolyPhen-2 | SIFT | LRT | CADD | GERP++ |
|----------------|-------------|------------------|---------------------|----------|------------|-------------|----------------|------------|------|-----|------|--------|
| <i>CARMIL1</i> | NM_017640.6 | c.3828_3847+1del | p.(Arg1277*)        | Het      | 0          | 0           | --             | --         | --   | --  | --   | --     |
| <i>SLC19A2</i> | NM_006996.3 | c.196G>T         | p.(Glu66*)          | Hom      | 6.31e-7    | 0           | D              | --         | --   | --  | 29.5 | C      |
| <i>SLC7A8</i>  | NM_012244.4 | c.550A>C         | p.(Thr184Pro)       | Het      | 1.4e-3     | 0           | D              | PrD        | D    | -   | 29.2 | C      |

Abbreviations: C, conserved; D, deleterious; het, heterozygous; hom, homozygous; PrD, probably damaging; T, tolerated