



A novel mutation in the TMC1 gene causes non-syndromic hearing loss in a Moroccan family

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Mots-clés	hearing loss [9], Morocco [10], TMC1; Mutation [11], Whole exome sequencing [12] Autosomal recessive non-syndromic hearing loss (ARNSHL) is one of the most common genetic diseases in human and is subject to important genetic heterogeneity, rendering molecular diagnosis difficult. Whole-exome sequencing is thus a powerful strategy for this purpose. After excluding GJB2 mutation and other common mutations associated with hearing loss in Morocco, whole-exome sequencing was performed to study the genetic causes of one sibling with ARSHNL in a consanguineous Moroccan family. After filtering data and Sanger sequencing validation, one novel pathogenic homozygous mutation c.1810C>G (p.Arg604Gly) was identified in TMC1, a gene reported to cause deafness in various populations. Thus, we identified here the first mutation in the TMC1 gene in the Moroccan population causing non-syndromic hearing loss.
Résumé en anglais	
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Liens

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