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A novel LHFPL5 mutation causes autosomal recessive hearing loss in an Omani family

Ahmed Al Amri
National Genetic Center , Royal Hospital, Oman

Statement of the Problem: Hearing loss (HL) is a debilitating disorder that impairs language acquisition, resulting in disability in children and potential isolation in adulthood. The condition could be seen as the only phenotype or with other comorbidities like cognitive impairment and depression. The onset of HL can have a genetic basis, though environmental factors which are often preventable can also cause the condition. The genetic forms are highly heterogeneous and early detection is necessary to arrange appropriate patient support. The purpose of this study is to identify the underlying genetic causes of hearing loss in selected consanguineous Omani families.

Methodology & Theoretical Orientation: Regions of shared homozygosity in the affected members of the studied families were determined using SNP array and homozygosity mapping technologies. Whole exome sequencing (WES) was applied to identify the list of candidate variants. Combining the data of homozygosity mapping and WES is a powerful strategy that allows finding the list of causative homozygous variants in these families. Various bioinformatics and genotyping methods were utilized in order to filter/select the most likely mutations. The refined causative variants were finally confirmed by segregation studies through Sanger sequencing.

Findings: The studied Omani family originated from first-cousin marriage and its pedigree suggested autosomal recessive/X-lined mode of inheritance. The applied methodology resulted in the identification of a novel mutation in LHFPL5 (p.L192P).

Conclusion & Significance: The family genetic studies confirms the likelihood of recessive disease (LHFPL5) but also provides some evidences that in such scenario, it is not always simple as compound diseases, multiple genes and an unexpected X-linked mutation could exist in a consanguineous family. The LHFPL5 mutation is likely to account for the deafness phenotype observed in this family, and this is beneficial to other family members via counseling and carrier screening. Affected children within the family can now be identified earlier and appropriate intervention offered, such as cochlear implants, to help with the child's speech and language development.

Biography

Ahmed Al Amri has experience working in next generation sequencing (NGS) technology where the research was done in families with hearing loss, intellectual disabilities and autistic spectrum disorder (ASD) and psychiatric disorders. He has gained his experience from Cardiff University, St George's, University of London (SGUL) and St James's University Hospital, Leeds. He is currently establishing whole exome sequencing (WES) at the National Genetic Center, Royal Hospital, Oman, as a service for different neurodevelopmental and complex conditions at the country.

ahmedamri@hotmail.com

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