10th International Congress on

STRUCTURAL BIOLOGY October 18-19, 2018 Helsinki, Finland

Prevalence of Non-syndromic hearing loss in UAE due to genetic mutation in Mitochondrial 12S rRNA

Maryam Alobathani University of Sharjah, UAE

Deafness is a disease of inability to hear referred as hear impairment or anacusis for those with low or no hearing. It's important at all levels of health care due to their significant burden on affected individuals and societies. Hearing loss can be classified as partial or completely inability to hear. Deafness can occur due to different mutations in different human genome such as Connexin26 the most common deafness gene, CDH23, TMC1 and in mitochondria. mitochondrial organelle is ubiquitous in eukaryotic as intracellular double membrane structure. The main function of mitochondria is to synthesis ATP by OXPHS and plays important roles in cell death and oxidative stress control. Mutation in mitochondrial DNA cause OXPHS defects that leads to many diseases one of them is hearing loss as syndromic and non-syndromic deafness. Therefore, we aim in this research to identify the genetic causes of NSAHL underlying wide spectrum of congenital conditions in UAE population. This study focused on a non-syndromic deafness caused by mutation on a mitochondrial MTRNR1 gene that encodes 12SrRNA. 40 Samples were collected from unrelated NSHL UAE family members and screened using biological tools such as PCR, Genomic sequencing machines and Bioinformatics tools. Expert geneticist recruiting and clinically assessing the participant patients involved in this study. The main approaches proposed are: (i) homozygosity maps were used to localize the homozygous regions in each particular family, (ii) next generation sequencing platform used to sequence the whole-exome of an affected individuals to explore all variants including the pathogenic mutations, finally (iii) pathogenicity of the outcoming variant results validated or ruled out by performing functional assays. The study indicates two out of the total 40 samples of unrelated UAE families with deafness due to mitochondrial 12S rRNA mutation m.669 T>C and m.827A>G, as well some known polymorphisms. This study is part of other researches done on a larger scale on similar area of interest that will reach to figure out new mutation related to mtDNA 12S rRNA variations. This finding can also help in future genetic counselling, prenatal screening, and postnatal genetic diagnosis to prevent the prevalence of more NSHL patients.

Biography

Maryam Alobathani has completed her Bachelor studies in Biotechnology by age 22 from University of Sharjah and know she's working as a laboratory supervisor and completing her Master studies at the same university.

Notes: