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Genetic mutations of mitochondrial 12s rRNA in non-syndromic hearing loss in UAE

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Deafness is the most predominant hereditary sensorineural disease worldwide. Mostly, congenital deafness is non-syndromic hearing loss (NSHL) cases with no association with other signs and symptoms mainly caused by damage in the inner ear from different types of genetic mutations. In this study, the focus was on the mutations in mitochondrial DNA 12s rRNA region that leads to NSHL in UAE. To study the molecular information of mtDNA 12s rRNA in affected individuals, a group of 39 unrelated UAE patients with NSHL were selected for mutational screening using PCR and DNA sequencing techniques. As for controls, 50 normal-hearing individuals were collected. Our study revealed the presence of some mitochondrial DNA 12s rRNA mutations previously reported with deafness m.669 T>C and m.827 A>G. We have detected also some known polymorphisms. In addition, new nucleotide variations were detected in UAE affected people. The study of their impact on the 12s rRNA structure suggested a pathogenic effect.

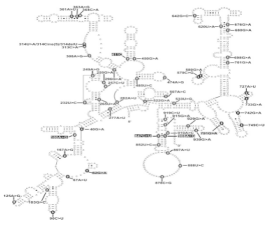

Figure-1: *Secondary structure map of the human 12S mt-rRNA showing sites of mutation.*

Recent Publications

1. A Almutery, W Kamal, H Alboushi, M Manjuran and N Rais (2017) Genetic polymorphism of vesicular monoamine transporter 1 gene (SLC18A1) in Emirati population, 6E9A99D64456. *International Journal of Genetics and Molecular Biology*; 9(3): 16-20.
2. H Al-Safar, W Kamal, A Hassoun, W Almahmeed and N Rais (2016) Combined association analysis of interleukin 1-receptor antagonist (IL-1RN) variable number of tandem repeat (VNTR) and Haptoglobin 1/2 polymorphisms with type 2 diabetes mellitus risk. *J Diabetes Metab Disord*; 15: 10.

Biography

Walaa Kamal is a PhD student in the Autonomous University of Barcelona and her major study focuses on genetics. She finished her Master degree in Human Genetics and Forensic Science from Manipal University, Dubai. She worked as a Researcher at Khalifa University, Abu Dhabi and has published three papers with other authors. Currently, she is an Assistant Researcher under deafness grant leading by Dr. Abdullah Al Mutery at University of Sharjah.

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