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Mutation spectrum of common deafness: Causing genes in patients with non syndromic deafness in UAE population

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Congenital genetic disorders are important at all levels of health care due to their significant burden on affected individuals and societies. They may be caused by genetic factors or be triggered by environmental exposures. High prevalence of consanguineous marriages is present in many communities throughout the world, especially countries of the Middle East and North Africa. Due to high consanguinity rates in these populations an increased rate of congenital monogenic disorders, particularly non syndromic autosomal recessive hearing loss (NSAHL) were noticed. This condition accounts for a substantial numbers of birth defects and disabilities among live births in UAE. Therefore, unravelling the genetic causes of NSAHLs is of great value for families and society as a whole. To provide accurate genetic testing and counseling in the UAE population, we investigated the molecular etiology of non syndromic deafness in UAE deaf population. Unrelated affected individuals with hearing impairment (n=60) were recruited for this study. Two common deafness-related genes, GJB2 and mtDNA 12SrRNA were analyzed using all exon sequencing. GJB2 mutations were detected in 15% (9/60) of the entire cohort. The mutation rate of mtDNA 12SrRNA in this group was 5% (3/60). These findings show the specificity of the common deaf gene mutation spectrum in UAE. According to this study, there were specific hotspot mutations in UAE deaf patients. Comprehensive sequencing analysis of the two common deaf genes can help portray the mutation spectrum and develop optimal testing strategies for deaf patients in UAE.

Biography

Tlili received his Ph.D. in biology engineering from National Engineering School of Sfax, Sfax university, Sfax-TUNISIA in 2007. During his Ph.D., he worked on genetic of deafness in Tunisian population. He spent two years as a Postdoctoral Fellow at the Pasteur institute (France). Tlili joined the Department of Applied Biology, University of Sharjah, Sharjah, UAE as a Full-time Assistant professor. He taught several courses in biology including Mendelian genetics, general biology, molecular biology, human molecular genetics and supervised several undergraduate and graduate students to conduct their research projects at his laboratory. Dr. Tlili has published several scientific papers through different research proposals granted as principal and associate investigator, in different abstracted, refereed and indexed Journals. Most of these papers were in the area of human molecular genetics. Also he has participated in several local, regional and international meetings to present his work as a talk or as a poster. Tlili is quite interested in working at a prestigious institute of high standards both in academic and research because this will give him the opportunity to establish a research network involving different laboratories in UAE and abroad.

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