World Academy of Science, Engineering and Technology
International Journal of Mathematical and Computational Sciences
Vol:14, No:12, 2020

Influence of Genetic Counseling in Family Dynamics in Patients with Deafness in Merida, Yucatán, Mexico

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Abstract : Hearing loss is an etiologically heterogeneous condition, where almost 60% is genetic in origin, 20% is due to environmental factors, and 20% have unknown causes. However, it is now known that the gene, GJB2, which encodes the connexin 26 protein, accounts for a large percentage of non-syndromic genetic hearing loss, and variants in this gene have been identified to be a common cause of hereditary hearing loss in many populations. The literature reports that the etiology in deafness helps improve family functioning but low-income countries this is difficult. Therefore, it is difficult to contribute the right of families to know about the genetic risk in future pregnancies as well as determining the certainty of being a carrier or affected. In order to assess the impact of genetic counseling and the functionality, 100 families with at least one child with profound hearing loss, were evaluated by specialists in audiology, clinical genetics and psychology. Targeted mutation analysis for one of the two known large deletions of upstream of GJB2/GJB6 gene (35delG; and including GJB2 regulatory sequences and GJB6) were performed in patients with diagnosis of non-syndromic hearing loss. Genetic counseling was given to all parents and primary caregivers, and APGAR family test was applied before and after the counseling. We analyzed a total of 300 members (children, parents) to determine the presence of the GJB2 gene mutation. Twelve patients (carriers and affected) were positive for the mutation, from 5 different families. The subsequent family APGAR testing and genetic counseling, showed that 14% perceived their families as functional, 62 % and 24 % moderately functional dysfunctional. This shows the importance of genetic counseling in the perception of family function that can directly impact the quality of life of these families.

Keywords : family dynamics, deafness, APGAR, counseling
Conference Title : ICSRD 2020 : International Conference on Scientific Research and Development
Conference Location : Chicago, United States
Conference Dates : December 12-13, 2020