

Role of genetic and biochemical factors associated with non syndromic hearing loss

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Introduction: Hearing impairment is one of the most common widespread birth defects and one of every 1000 new born has bilateral permanent Sensorineural Hearing Loss (SNHL). About 50% of SNHL involves genetic factors and 46 genes have been identified as causally related to nonsyndromic SNHL. In spite of this large genetic heterogeneity, mutations in *GJB2* and *GJB6* genes are primarily responsible for most of the genetic hearing loss. A free radical is an unstable cluster of molecules can cause errors in genetic “messages” by altering DNA. This can, among other things, lead to a reduced blood supply to organs such as the inner ear and brain, there by damaging hearing. Our bodies produce enzymes known as antioxidants, such as Superoxide Dismutase (SOD) and catalase to counteract the damage.

Objectives: To study to investigate the genetic cause of deafness by genetic screening of the *GJB2* genes and enzyme and catalase in deaf patients.

Methodology: Patients with sensorineural congenital HI were selected after obtaining their consents. Clinical samples were tested using PCR/RFLP for 35delG & 167delT mutations. PCR was used to amplify two regions of the exon 2 of connexin26 and PCR products were analyzed using *Bsl* I and *Pst* I. Blood level of SOD and catalase was estimated by spectrophotometric analysis.

Results: 87.8% had normal genotype for the studied mutations, 4.88% were compound heterozygotes, 7.32% homozygotic for the 35delG mutation, 9.76% heterozygotic for 35delG/*GJB2*. The 167delT mutation was not detected in any of the deaf individuals.

Discussion: Our data confirmed the presence of the 35delG mutation in the *GJB2* gene in cases of non-syndromic bilateral moderate to profound sensorineural hearing loss in West Bengal. For the 167delT mutation the allele frequency was zero for the familial and sporadic groups. These findings underline the importance of a genetic diagnosis that may clarify the etiology and provide early treatment for children. Genetic counseling for their family members will help in early detection and treatment.

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