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New Mutation Haplotypes in Non-Syndromic Hearing Loss

We report on a male Caucasian child of two years of age, who attended Early Intervention Clinic with history of speech delay. There was family history of hearing loss in paternal uncles and aunts. Physical examination was normal with appropriate development and non-dysmorphic features. Assessment of internal, middle and external ears revealed no abnormality. Pure tone audiometry revealed bilateral profound sensorineural hearing loss of more than 90db of both middle and high frequency sounds. Genetic study was performed by PCR sequencing of the DNA carrying coding exon 2 of the gene *GJB2* (*Connexin 26*) and its flanking sequences (235nt). It identified c.35delG and c.427delC mutations in the aforesaid gene. Analysis of electrophoretogram revealed these two mutations in *Cis* position which has never been previously described in literature. He was subsequently fitted with hearing aids and also underwent hearing rehabilitation (speech therapy). At three years of age, he showed good improvement in his speech and had started using two to three word sentences.

Almost 80% of hereditary hearing loss is of nonsyndromic type [1]. Autosomal recessive, non-syndromic hearing loss and deafness (DFNB1) is characterised by non-progressive, mild to profound congenital hearing impairment that is detected at an early age. Though genetic linkage has demonstrated more than twenty hitherto unidentified genes in its causation, allelic mutation at *GJB2* locus has been reported in more than 90 percent of deafness cases [1,2]. Single nucleotide deletion of a Guanine residue from string of 6 guanine nucleotides from position 30 to 35 (c.del30G or c.del35G) is the most common identified mutation in Caucasian patients. Clinical heterogeneity of GJB2 mutations support the role of other unidentified genetic factors and environmental effects [3].

Assignments of alleles to the chromosomes (Haplotype) yields important information regarding recombination. Recombination events are marked in both family- and population-based linkage analysis to pinpoint disease causing mutations. Often haplotypes (several *cis* acting sites) are required to produce a phenotype, affecting gene products. Haplotypes by affecting gene product can either increase or decrease the severity of the phenotype [4]. Though identification of c.427delC as a *Cis* position haplotype could not be attributed to have any effect on phenotypic expression in our case as c.35delG alone has the potential to cause mild to profound hearing loss, similar reporting of further case or series with phenotypic attributes of this *cis* position haplotypes would help formulating the particular role of this haplotype (c.del35G and c.427delC) in modifying clinical expression of congenital non-syndromic hearing loss. We hope this case highlights the importance of genetic testing in children with sensorineural deafness as it helped us to pick up this novel haplotype which has not been previously reported in medical literature.

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