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Founder effect analysis of disease haplotypes in DFNB23/ USH1F linked pakistani families

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Usher syndromes are a group of autosomal recessive disorders characterized by moderate to profound sensorineural hearing loss and progressive visual loss from retinitis pigmentosa. Clinically they are classified into three types on the basis of phenotypes. Within each clinical group molecular heterogeneity exists and people with indistinguishable phenotypes have mutations in different genes. Protocadherin-15 (PCDH15) is one of the five genes identified as being mutated in Usher 1 syndrome and defines Usher syndrome type 1F (USH1F). Mutation in this gene also causes nonsyndromic deafness DFNB23. A total of 25 families were collected in which pattern of inheritance was autosomal recessive and were screened for locus DFNB23 by using fluorescently labeled markers D10S2529, D10S546, and D10S2522. Three families were found to be linked with DFNB23. Haplotypes of these families were compared with 12 previously linked families obtained from CEMB repository. Seven families divided into two groups shared same haplotypes while in other eight families, no correlation was found between the haplotypes. Variability of haplotypes among families indicates presence of different type of mutations and families with same haplotypes may have same founder. These results will lead to better understanding of hearing impairment caused by mutations in PCDH15 and will help in identification of carriers and genetic counseling.

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