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Prevalence of Cx26 (GJB2) Gene Mutations Causing Recessive Nonsyndromic Hearing Impairment in India

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ABSTRACT Autosomal Recessive Nonsyndromic Hearing Impairment (ARNSHI) is caused by mutations in the gap junction gene GJB2 or Connexin26 gene (Cx26) in many of the world populations. In the present study screening of 200 probands with profound Nonsyndromic Hearing Impairment (NSHI) in comparison to 200 normal hearing controls from Andhra Pradesh, India revealed high prevalence of W24X mutation (6.5%) and low frequency of W77X (0.5%) and 235delC (0.5%) mutations. Incidence of 35delG and 167delT were not detected. High incidence of heterozygosity with R127H both in NSHI patients (28.0%) and in controls (36.5%) were observed indicating that the mutation could be a polymorphism and may not be the cause of NSHI. Two of the probands (1.0%) showed homozygosity for the mutation and causative nature of this mutation has to be evaluated. Cx26 mutations causing hearing impairment are found to be specific to certain populations exhibiting ethnic diversity.