عنوان مقاله:

Relative Frequency of ۳۵delG Mutation in GJB۲ Gene in Autosomal Recessive Non-Syndromic Hearing Loss (ARNSHL) Patients in Kerman Population

محل انتشار:

مجله دانشگاه علوم پزشکی کرمان, دوره 11, شماره 3 (سال: 1383)

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خلاصه مقاله:

Congenital hearing loss with many genetic and environmental causes affects ۱ in ۱۰۰۰ newborns. Mutations in the GJB۲(Gap Junction Beta-۲) gene encoding the gap junction protein connexin ۲۶ have been established as the main cause of autosomal recessive non-syndromic hearing loss. The aim of this study was to study the frequency of one mutation (۳۵delG) of GJB۲ gene in Kerman non-syndromic deaf population. For this purpose, ۱۳۰ chromosomes from ۶۵ patients were studied and ۳۵delG mutation was diagnosed in ۳(۲.۳%) chromosomes (one patient was homozygote and the other one was .heterozygote). This rate of frequency is significantly higher comparing to that in the whole population of Iran

کلمات کلیدی:
Autosomal recessive non-syndromic hearing loss (ARNSHL), GJB۲, ۳۵delG