عنوان مقاله:

Profile of Iranian GJB۲ Mutations in Young Population with Novel Mutation

محل انتشار:

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خلاصه مقاله:

Objective(s) Despite the enormous heterogeneity of genetic hearing loss, most non-syndromic hearing losses are caused by mutations in the GJB۲ gene. We aimed to characterize the mutation profiles of ۱۰۰ Iranian deaf patients that were under ۱۰ years old. Materials and Methods Patients were tested with direct sequencing of entire coding region of the GJB۲ gene. Results Eight known mutations plus one novel (۳۵۸delGAG) were found in ۲۵% of study group. The ۳۵delG mutation (۶۴%) constituted the majority of GJB۲ mutations. Conclusion Role of GJB۲ mutation in Iranian young deaf population is more prominent than previous study that can be a result of higher consanguine marriage in population. But our result shows that there is only ۲۵% non- syndromic hearing loss due to high frequency of consanguine marriage in Iranian population. Identification of other genes involved in genetic deafness will help us understand the fundamental mechanisms of normal hearing, both in early diagnosis and therapy.

کلمات کلیدی:

ARNSHL, Connexin Cx۲۶, GJB۲, Hereditary hearing loss