عنوان مقاله:

Carrier frequency of p.R۳۶۸H in glaucoma causing gene CYP۱B۱ may justify pre-marital screenings in eastern Guilan

محل انتشار:

چهارمین کنگره بین المللی و شانزدهمین کنگره ملی ژنتیک (سال: 1399)

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خلاصه مقاله:

Background and Aim: Glaucoma is a major cause of blindness. Primary congenital glaucoma (PCG) is the most severe form of glaucoma. Genetic factors significantly contribute to its etiology, and CYP۱B۱ is its most important causative gene. In the first study on the genetics of PCG among Iranians, it was shown that mutations in CYP۱B۱ were the cause of disease in approximately ۷۰% of ۱۰۰ patients studied. Also, among ۱۹ different mutations identified, four were most frequent. Additionally, the western and north western regions of Iran had the highest prevalence of PCG, and the distribution of the mutations in various regions of Iran differed. Guilan was a province with high PCG prevalence. A subsequent study on ۷۰۰ patients from Guilan, suggested that mutated alleles that are causative of p.G۶۱E and p.R۳۶۸H were frequent specifically in, respectively, Talesh and eastern regions of Guilan. Here, we aimed to get a more accurate estimate of carrier frequencies of these mutations in these regions in order to consider need for pre-marital screenings.Methods: In order to achieve a carrier frequency estimate with maximum error of ۲%, ۱۰۰۰ individuals from Talesh and ۳۰۰۰ from eastern regions of Guilan needed to be screened for the respective mutations. Individuals were recruited based on cluster sampling. DNA was extracted from saliva samples. The c.۱۸۲G>A causative mutation of p.G۶۱E was screened in the Talesh samples using an RFLP protocol. The c.۱۱۰۳G>A mutation causative of p.R۳۶۸H was screened using an ARMS PCR protocol. Statistical analyses were done using OpenEpi.Results: Nine individuals among ۱۰۳۶ from Talesh were shown to be carriers of the p.G۶۱E mutation, and ۷۳ among ۳۰۲۹ individuals from eastern regions of Guilan were carriers of the p.R۳۶۸H mutation. These figures indicate a carrier frequency of ۰.۰۰۸۶ (۹۵% confidence interval: ۰.۰۰۴۵ – ۰.۰۱۶۴) for p.G۶۱E in Talesh, and ۰.۰۲۴ (۹۵% confidence interval: ۰.۰۱۹ – ۰.۰۳۰) for p.R۳۶۸H in east of Guilan.Conclusion: The carrier frequencies calculated are within ranges previously assessed, but with significantly improved confidence intervals. Based on results of premarital screenings of thalassemia causing mutations on incidence of this disease in Iran, premarital screenings of the p.R۳۶۸H may be justified.

کلمات کلیدی:
primary congenital glaucoma, CYP۱B۱, p.R۳۶۸H, eastern Guilan, Carrier frequency