

Predisposing Mutations of *β-Myosin Heavy Chain* gene in Iranian Azeri Patients with Hypertrophic cardiomyopathy

Abstract

Background: Hypertrophic cardiomyopathy (HCM) is a serious lethal disease that is characterized through involvement of heart muscles and cause sudden death. HCM is known to be manifested by mutation in at least 12 sarcomeric genes. The *β-Myosin Heavy Chain (β-MYH7)* gene mutations are the most common gene with mutations in HCM. In the current study, it is aimed to identify mutations of *β-MYH7* gene.

Method: Forty patients with HCM of Azerbaijan, Iran race without any familial relations were included in ongoing study. Genomic DNA content was extracted from blood samples using Phenol-Chloroform method. Then, all the exonic regions of MYH7 were amplified by PCR. Afterwards, the PCR products were screened by single strand conformation polymorphism (SSCP) to identify the mutated positions. SSCP results were approved by sequencing the amplified exons.

Results: Thoroughly, 7 patients were discriminated to bear 7 mutations in *β-MYH7* gene which all of them were missense mutations. There were no novel mutation among patients and all mutation that was realized in this study had been explained before. There were 2 patients with G12742a, 2 with G12771a, and three patients harbored T8772c, T12315c, G13248a mutations. The mutations identified in the present study are listed in Table 3 with more details.

Conclusion: This study concluded that *β-MYH7* gene mutations were presented in only 18 percent of the patients. However other similar studies had already resulted in at least with 25 percent frequency. The discrepancies are perhaps because of the technics and differences in races.

Keywords: HCM, β -MYH7, SSCP-PCR