عنوان مقاله:
Genetic study of hypertrophic cardiomyopathy

محل انتشار:
سومین کنگره بین المللی پزشکی شخصی ایران (سال: 1397)

تعداد صفحات اصل مقاله: 1

نویسندگان:
Elnaz Abedini - *۱۹ mayıs univercity /Samsun /Turkey*

Mohammad Asadi - *Shahid Beheshty univercity/Tehran / Iran*

خلاصه مقاله:
Hypertrophic cardiomyopathy (HCM) is the most common inherited cardiovascular disease. HCM is a highly complex and heterogeneous disease regarding not only the number of associated mutations but also the severity of phenotype, symptom burden, and the risk of complications, such as heart failure and sudden death. The penetrance is incomplete and it is age and gender dependent. It is accepted as a disease of the sarcomere. Sixty percent of HCM cases carry mutations in 1 of 8 sarcomere protein genes, mainly non-sense MYBPC3 and missense MYH7 variants. Young patients with severe phenotype and other clinical features are included in proposed scores for prediction of high positive genetic result. The number of genes reported as disease-causing has increased in the last few years, in some cases without robust evidence. Currently available in silico tools are not always useful for differentiation between benign and deleterious variants. There is enough information on genotypephenotype correlations to start understanding the mechanisms of the disease. Genetic and environmental modifiers have been explored with some interesting insights from miRNA studies with potential as biomarkers and therapeutic agents. There is an additional value of genetic testing in HCM for prognosis. Knowledge about genetics and functional studies are the basis of near .future therapies

کلمات کلیدی:
Hypertrophic cardiomyopathy، Genetic ، MYBPC3، MYH7