عنوان مقاله:

Genotype-phenotype correlation and risk assessment in patients with diagnosis Brugada Syndrome

محل انتشار:

اولین کنگره پزشکی شخصی (سال: 1395)

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خلاصه مقاله:

Introduction:Brugada syndrome (BrS) is an autosomal dominant inherited channelopathies characterized by ST-segment elevation in V1-V2 leads followed by negative T-wave on standard ECG, and high risk of sudden cardiac death (SCD). The disease was considered of the high frequency in Southeast Asia, but current estimation of BrS is at least 1:10000 in all ethnic groups. Approximately 15-30% of individuals with BrS cases are affected by loss of function mutations in SCN5A gene (more than 370 mutations). In this study, we study genotype-phenotype correlation and risk stratification in our group of patients.Materials and methods:The present study was performed in accordance with the Helsinki Declaration and local ethics committee. Written informed consent for clinical and genetic evaluation was obtained from each member.The clinical follow-up included a review of personal and familial history, complete physical examination, 12-lead ECG, 24-hour Holter ECG monitoring, echocardiography, and sodium channel blocker challenge test. Genetic analyzing of SCN5A gene was performed.Results:Age of first detection of Brugada syndrome is 37±13 years old, male/female ratio is 8:1. In our group, the most common symptoms have been a history of SCD cases in the family (56%), syncope (51%). Brugada Pattern type 1 detected in 44% of our group, in superficial 12-leads ECG. Ventricular tachycardia was detected in 32% of patients. We had found 17 mutations in SCN5A gene in 17 unrelated probands (21%). 11 of 17 mutations (65%) are reported as first time. Functional study was performed for all new mutations by In-Silico and cellular electrophysiological study was done for one of them.All mutations were found in Male gender. Regards to our SCN5A gene analysis, around 50% of mutations are missense and in half of them had recorded not any history of life-treating symptoms. Conclusion:Correlation between Syncope and positive familial history of SCD under 45 years old in BrS patients and SCN5A mutations were found ($P<0.01$).ECG study in our group was shown strongly correlation between SCN5A gene mutations and prolonged PR interval (PR> 200msec). Regards to at least 3 years follow up, Prognosis in BrS patients who carrier of haploinsufficiency mutations in SCN5A gene are poor compare with missense mutations ($p<0.05$). ICD implantation is recommended in patient with mutation in SCN5A gene.It seems, our data on the role of SCN5A gene study help us not only to BrS diagnostics but also in prognosis for patients.

کلمات کلیدی:
SCN5A gene ,Brugada syndrome, Sudden Cardiac Death