ABSTRACTS

ASSOCIATION BETWEEN hRYR2 MUTATIONS WITH VENTRICULAR TACHYCARDIA IN KAZAKH POPULATION

Zh.Abilova[1], M.Bekbossynova[2], A.Akilzhanova[1]

Laboratory of Genomic and Personalized Medicine, National Laboratory Astana, Nazarbayev University (Astana, Kazakhstan)
[2]JSC National Research Cardiac Surgery Center (Astana, Kazakhstan)

Key words: Arrhythmia, ryanodine receptor, sequencing, ventricular tachycardia, mutation

Introduction: Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an inherited disease characterized by polymorphic or bidirectional ventricular arrhythmias (VA) triggered by physical or emotional stress in young people with a structurally normal heart.

Materials and methods: Patients with arrhythmia - ventricular tachycardia were involved into research. Diagnosis was verified in National Research Cardiac Surgery Center, Astana. Target areas of hRYR2 gene including the most important 45 exons were amplified with PCR and directly sequenced.

Results: Genetic variants of hRYR2 gene in two catecholaminergic polymorphic ventricular tachycardia (CPVT) patients and 14 ventricular tachycardia (VT) patients were screened. Total number of VT patients whose mutation in hRYR2 gene screened was 35 people. Also genetic analysis was carried out for relatives of patient who had been observed to carry mutation in hRYR2 gene. New mutations in CPVT patients *(c.A13892T; p.D4631V)* and new mutation in one VT patient (c. *G5428C;p.V1810L)* was detected. Both variants are located in phylogenic conservative areas of hRYR2 gene and seems to be pathological according to MutationTaster and PolyPhenII prediction programs. Also three well-known synonymous polymorphisms *rs3765097, rs2253273* and *TMPESP 1237 664 067* in studied group were detected. Moreover mutation *(c.C7511T; p.T2504M)* was found which previously was detected in arrhythmogenic right ventricular dysplasia patient. This variant is located in phylogenic conservative areas of hRYR2 gene and assessed as pathological (points by *MutationTcister D* (0.99) and by *PolyPhenllD* (0.99)).

Conclusion. This research is useful to evaluate necessity of genetic screening and reliable genetic consultation for ventricular rhythm disorder patients in order to predict and prevent sudden cardiac death.