

Genetic counseling of ventricular tachycardias

To the Editor,

I read with interest the article, "Clinical spectrum and long-term course of sustained ventricular tachycardia in pediatric patients: 10 years of experience" by Şengül et al. (1). However, I did not find any mention of genetic evaluation in these patients, especially in the 14 patients with sudden cardiac deaths in their families at a young age. In addition, 65.9% of the patients were grouped as having "idiopathic VT." Did they have any genetic testing?

Inherited cardiac arrhythmias [long QT syndrome, short QT syndrome, catecholaminergic polymorphic ventricular tachycardia (CPVT), and Brugada syndrome] give rise to ventricular tachycardias, and these rare diseases are often the underlying cause of sudden cardiac death in young individuals. Mutations in several genes encoding ion channels or proteins involved in their regulation are often the underlying cause and can now be tested. For example, in CPVT, the *RYR2* gene mutations have autosomal dominant inheritance, and *CASQ2* and *TECRL* have autosomal recessive inheritance pattern (2).

As more patients with congenital heart diseases reach adulthood and have children, it is of utmost importance that they have genetic counseling. Genetic counseling can also help in diagnosis and managing the course of disease.

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Author's Reply



To the Editor,

We would like to thank the authors for their interest in our manuscript "Clinical spectrum and long-term course of sustained ventricular tachycardia in pediatric patients: 10 years of experience" (1) and for taking the time to express their concerns.

Ventricular tachycardias (VT) are potentially life-threatening arrhythmias that can cause sudden cardiac death. Inherited cardiac arrhythmias and cardiomyopathies are also the underlying cause of sudden cardiac death in young individuals (2-4). As you mentioned in the article titled, "Genetic counseling of ventricular tachycardias" we agree that genetic counseling has a crucial role in the diagnosis and management of inherited arrhythmias/cardiomyopathies. However, the primary objective of our study was to define the clinical aspects of pediatric patients who were diagnosed as having sustained VT. In our study, patients with cardiomyopathies, catecholaminergic polymorphic VT (CPVT), and patients without structural heart disease were included; however, patients with Brugada syndrome and those who developed ventricular fibrillation as long QT syndrome were excluded. Therefore, we did not mention the genetic results of the patients in our article.

We appreciate their acknowledgment of the genetic screening of patients with such life-threatening arrhythmias. The genetic test results of patients diagnosed with cardiomyopathy in our study were as follows: PKP2 (n=1), RYR2 (n=1), and DSG-2 (n=1) gene mutations and normal genetic screening (n=2) were detected in patients with arrhythmogenic right ventricular dysplasia (ARVD/C) (5). ACTN2 (n=1) and TTN (n=2) gene mutations were detected in patients with left ventricular noncompaction cardiomyopathy, and MYH7 (n=2) and MYBPC3 (n=2) gene mutations were detected in patients with hypertrophic cardiomyopathy (6). In addition, RYR2 (n=6), CASQ (n=4), TRDN (n=2), and KCNJ2 (n=1) gene mutations were detected in patients diagnosed with CPVT. Actually, if there is no suspicion of any cardiomyopathy (especially ARVD/C), we do not routinely screen for genetic mutations in patients with idiopathic VT. In this study, we had a confusing case in the idiopathic VT group of an infant who had been previously treated at our clinic with radiofrequency catheter ablation for multidrug-resistant, recurrent VT episodes; and he had had three more sessions of ablations that were performed at another center; however, he deceased suddenly at home shortly thereafter. We could not perform cardiac MRI and genetic screening in this case. As a developing country, as genetic studies could not be performed routinely in our clinic before 2015, the patients who were diagnosed before then do not have genetic results. However, after 2015, genetic studies were performed on patients suspected clinically.

Ultimately, we agree with the authors that patients with life-threatening arrhythmias need genetic counseling for accurate diagnosis and management of the disease.

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