Abstract
Following a brief loss of consciousness during an athletic activity, a 41-year old male was found to have a genetic abnormality of heart rhythm control, catecholaminergic polymorphic ventricular tachycardia (CPVT). The condition was treated with an implantable defibrillator and drug therapy with beta blockers. Additional antiarrhythmic drug therapy with Flecainide is being considered. Genetic testing has been advised to allow definitive screening of family members.
CASE STUDY
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)

Patient Presentation
• A 41-year old male track and field athlete suffered a brief loss of consciousness while running hurdles.
• On careful questioning, the patient related a history of palpitations with dizziness associated with high-level exercise or emotional stress since adolescence.

Assessment
• An exercise treadmill test showed multiple long bursts of rapid ventricular tachycardia during exercise.
• An echocardiogram showed normal right and left ventricular ejection fractions, with mild RV dilatation and mild concentric left ventricular hypertrophy, consistent with an athlete’s heart.
• Patient underwent cardiac catheterization, which showed normal coronary arteries.
• A cardiac MRI showed no areas of scarring, and no evidence of hypertrophic cardiomyopathy or arrhythmogenic right ventricular dysplasia.

Diagnosis
• Based on the details of the patient history and the heart-rate dependency and morphology of the ventricular tachycardia, the diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) was made.

Treatment
• An implantable defibrillator was placed over the left pectoral muscle for prevention of sudden death.
• The patient was initially treated with metoprolol with dose titration guided by exercise treadmill. He continued to have episodes of ventricular tachycardia, and ultimately was switched to nadolol, which has demonstrated greater efficacy for CPVT.
• Genetic testing was recommended to permit definitive screening for the patient’s children and other family members. Currently, causative mutations can be identified in about half of patients with CPVT.

Outcomes
• The patient continues to exercise with limitations recommended. He has experienced appropriate defibrillator shocks for ventricular tachycardia leading to ventricular fibrillation associated with exercise or emotional stress.
• The role of flecainide therapy has been discussed as this drug has demonstrated unique efficacy for preventing ventricular tachycardia in patients with the CPVT mutation. This may allow the patient to reduce his beta blocker dose, which is his personal preference.
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**Conclusion**

- Patients with aborted sudden death must be carefully screened not only for the most common disorders such as coronary artery disease, but also for inherited disorders such as CPVT.
- Genetic testing and genetic counseling play a crucial role in proper management of these patients and their families.

“Because a variety of factors influence the type and severity of cardiac disorders, it’s essential that we learn as much as possible about the patient’s personal and family medical history, lifestyle, and other potential contributors to his or her condition. Making the correct diagnosis allows us to go beyond simply recommending defibrillator implantation, and offer specific therapy for these unique entities.”

– Susan O’Donoghue, MD
Individuals with a known inherited arrhythmia or a family history of sudden cardiac death may benefit from cardiogenetic evaluation. If you would like to discuss a patient, Dr. O’Donoghue can be reached at 202-877-7685, or call 202-877 GENE(4364) to leave information.

To learn more, please visit MedStarHeartInstitute.org.