A 28-year-old medical resident and marathon runner was admitted to the cardiology unit after experiencing cardiac arrest while jogging. On admission, she was noted to have a prolonged QT interval and a low potassium level (2.4 mmol/L). However, no signs of ischemia, prior arrhythmias (such as ventricular fibrillation), prior syncopal episodes or other cardiac findings were noted on history or ECG (Figure 1). An echocardiogram during subsequent workup documented normal left ventricular size and systolic function. A CT scan revealed slight right ventricular abnormality; however, this finding was attributed to her strenuous exercise routine and therefore ignored. A diagnosis of prolonged QT syndrome aggravated by hypokalemia was made, and the patient was advised to undergo implantation of a cardioverter defibrillator (ICD).

Several weeks later, the patient underwent another ECG, which showed no evidence of a prolonged QT interval and was normal at 448 millisec. Her PR interval was also normal as was her heart rate of 66 beats per minute.

Further evaluation over the following months and several consecutive ECGs that were negative for QT prolongation led to the removal of the long QT syndrome diagnosis and consideration of the possibility of atypical catecholaminergic polymorphic ventricular tachycardia (CPVT), especially because of the patient’s strenuous exercise regime. Upon CPVT testing (resting ECG, stress test and genetic testing), however, the patient showed no signs of the condition. Because of the patient’s lack of symptoms and maximal protection with an ICD, she was advised to continue her exercise regimen, which was an important part of her life.

Approximately five years later, the patient was admitted to the hospital because of increasing shocks and cardiac events occurring both during exercise and at rest. She underwent another ECG, which showed T-wave inversion and premature ventricular complexes (PVCs) (Figure 2), as well as a chest CT scan (Figure 3), which indicated right ventricular enlargement, fibrofatty change and apical hypokinesis, all of which raised the possibility of arrhythmogenic right ventricular cardiomyopathy (ARVC). Genetic studies confirmed the patient’s diagnosis of genetic (PKP2-positive) ARVC.

Discussion
Diagnosis of cardiac events in young athletes can be challenging. In any differential for such patients, we must consider several possibilities (left ventricular hypertrophy, right ventricular arrhythmias, cardiomyopathies, long QT syndrome, sinus bradycardia, atrial fibrillation, valvular heart disease and coronary disease) in order to prevent misdiagnosis and downstream complications.

Arrhythmogenic right ventricular cardiomyopathy is a rare cardiomyopathy that can lead to sudden cardiac death. It is caused by a combination of genetic (ie, desmosome mutation) and/or acquired environmental factors (ie, exercise). Diagnosis of ARVC remains a challenge because of its heterogeneous clinical presentation as well as its variable genetic expressivity and penetrance. In fact, only 30% to 50% of cases of ARVC reported show evidence of family history.
Practical ways of distinguishing ARVC from other cardiac conditions that have similar presentation (ie, long QT syndrome and CPVT) involve a combination of the following: 1) ECG, 2) right ventricular angiography/CT scan and 3) genetic testing. Although an ECG can show prolongation of the QT interval in patients with and without long QT syndrome, the QT interval is consistently prolonged only in those patients with a diagnosis of long QT syndrome. Second, a key diagnostic tool for ARVC is the use of right ventricular angiography or CT scan to detect an akinetic or dyskinetic subtricuspid, apical or infundibular right ventricle. In a young patient, this finding is characteristic for ARVC and unlikely for long QT syndrome or CPVT. Third, although genetic testing is not the most sensitive test for ARVC, testing of common mutations that can provoke ARVC (ie, PKP2/plakophilin-2) is likely to aid in excluding other diagnoses. Although our patient did have a temporarily prolonged QT interval and a cardiac event, there was no evidence of genetic long QT syndrome or a consistently prolonged QT interval, thus her diagnosis of long QT syndrome was appropriately rescinded.

Treatment of ARVC can be equally difficult. Although current therapeutic options include beta blockers, antiarrhythmic drugs, catheter ablation and implantable cardioverter defibrillator (ICD), lifestyle modifications may be as, if not more, important in treatment of these patients. In fact, studies conducted by La Gerche et al. have found that endurance exercise such as marathon running can induce right ventricular dysfunction and potentially worsen ARVC.

In this case, even though the patient was maximally protected from a cardiac event with an ICD and had a healthy lifestyle, she still suffered from subsequent cardiac deterioration, which is likely the result of her strenuous exercise plan coupled with her genetics.

This case teaches us that we must consider the patient’s whole story before diagnosing and treating. Lifestyle practices such as exercise, which is typically encouraged by the medical community, must be prescribed carefully and individualized to the patient prior to implementation; otherwise, patients may suffer from preventable complications.

Figure 1. ECG upon admission (2007). Note sinus rhythm and low-voltage QRS.

Figure 2. ECG upon follow-up (2012). Note sinus rhythm, low-voltage QRS, T-wave inversion, premature ventricular complexes and T-wave abnormalities (suggestive of inferior and anterior ischemia).

Figure 3. CT scan showing stable RV dilatation, reduced function and myocardial fatty deposition consistent with ARVC (2012).

REFERENCES


