Dynamic QT Changes in Long QT Syndrome Type 8

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Gain-of-function CACNA1C mutations have been shown to cause long QT syndrome type 8 (LQT8), an inherited arrhythmia with a marked QT prolongation and a bizarre form of polymorphic ventricular tachycardia (torsade de pointes) on electrocardiogram (ECG). Resting as well as dynamic ECG change is an important diagnostic hallmark for long QT syndrome, but the dynamic feature has not been previously reported in LQT8. The index patient was a 7-year-old asymptomatic boy referred to hospital because of abnormal ECG findings at a school health checkup (Figure A). The 12-lead ECG at rest showed a QT prolongation and a late-onset T wave. Genetic testing identified a heterozygous de novo CACNA1C mutation (p.Arg518Cys; R518C; Figure B, C), confirming the diagnosis of LQT8. Exercise stress test showed a transient normalization of a corrected QT interval (QTc) at peak exercise. The QTc prolongation returned to that of the control level before exercise at 4 min of recovery (Figure D). Similar ECG features at rest have been reported in LQT type 3 (LQT3). Similarly, in LQT3 patients, exercise tolerance test shortened QTc at peak exercise. Thus, dynamic and resting ECG features in the present LQT8 case mimicked those reported in LQT3, suggesting a similarity in that both result from gain-of-function mutations in genes encoding cardiac inward currents.

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