Hypertrophic Cardiomyopathy and Long QT Syndrome, Risk of Sudden Cardiac Death, and Importance of Genetic Testing: A Case Report

Background: Hypertrophic Cardiomyopathy (HCM) is the most common inherited cardiomyopathy and exhibits autosomal dominant pattern (1). Although, long QT has been reported in about one in eight patients with HCM (2), Long QT syndrome (LQTS) incidence is still unknown. The re-polarization abnormalities related to the hypertrophied myocardium in HCM patients can result in QT changes and the predisposition to malignant arrhythmias. The presence of LQTS in addition to HCM, could be a marker for higher risk of ventricular arrhythmia and an important screening tool for risk of sudden cardiac death (SCD) in these patients.
We present one such case of a patient with HCM and LQTS and strong family history of SCD.

Case: Our patient is a 44-year-old male who initially presented to the ER with progressive shortness of breath and fatigue. Echocardiography revealed severe concentric left ventricular hypertrophy and systolic anterior motion of the mitral valve leaflet. Significant left ventricular outflow (LVOT) obstruction was noted at rest. The peak velocity across the LVOT was 5.33cm/s with a peak pressure gradient of 114 mmHg. Moderate mitral regurgitation was noted. Because of the new diagnosis of HCM, cardiac magnetic resonance imaging (MRI) was obtained. This revealed significant amount of late gadolinium enhancement (LGE) in the mid infero-septal wall with end-diastolic wall thickness of 30 mm.
Family history was positive for HCM in one brother (already has a defibrillator) and another brother who died suddenly at age of 50 years while sleeping. In addition to left ventricular hypertrophy, his ECG showed prolonged corrected QT interval of 543msec.
He was started on guideline directed medical therapy. Outpatient external prolonged cardiac monitoring showed episodes of non-sustained ventricular arrhythmia with rates up to 136 beats per minute, lasting up to 14.1 seconds. Genetic testing revealed one pathogenic variant KCNQ1 associated with the arrhythmia condition of LQTS type 1. His calculated American Heart Association risk of sudden cardiac death was 13.4% in 5 years. Subcutaneous defibrillator was implanted for primary prevention of SCD.

Conclusion: HCM is the most common inherited cardiomyopathy. The association of HCM with long QT interval is known but the incidence of LQTS in these patients is not clear. This case report shows the importance of genetic screening in these patients. Identifying the additional defect could be crucial for patients and their family members' SCD risk stratification

REFERENCES:

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