Sudden Death in an Infant With Angina, Restrictive Cardiomyopathy, and Coronary Artery Bridging

An Unusual Phenotype for a β-Myosin Heavy Chain (MYH7) Sarcomeric Protein Mutation

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The patient, a female infant aged 9 months, presented to a community hospital emergency room with fever and was referred for cardiac evaluation because of an abnormal chest radiograph. Her ECG showed biatrial enlargement and inferolateral ST depression. An echocardiogram showed a dilated left atrium and a mildly dilated and thickened left ventricle. Systolic function was preserved but diastolic filling showed restrictive physiology. A clinical diagnosis of restrictive cardiomyopathy (RCM) with associated myocardial hypertrophy was made.

The patient underwent a comprehensive metabolic screen, as well as genetic testing (Hypertrophic Cardiomyopathy [HCM] Panel, Laboratory for Molecular Medicine, Cambridge, MA), which revealed a mutation (A1157G) in exon 13 of the β-myosin heavy chain (MYH7) gene, resulting in a nonsynonymous amino acid change from tyrosine to cysteine at position 386. This mutation has not been reported in the literature but had been seen previously by the Laboratory in an infant with de novo HCM. The mutation was categorized as being likely pathogenic.

The patient was asymptomatic, and she was started on metoprolol to limit her maximal heart rate and acetylsalicylic acid for thromboprophylaxis. At 16 months of age, the patient presented with episodes of grunting and crying, with generalized discomfort and diaphoresis. A Holter monitor showed significant, rate-related ST depression with an increase in troponin T to 0.01 μg/L (normal <0.01 μg/L). These episodes were presumed to be related to myocardial ischemia, and the child was treated with oral morphine, increased metoprolol, and was listed for cardiac transplantation. At 18 months of age, the child became ill with vomiting, lethargy, and diaphoresis and presented to a local emergency room with absent vital signs. The ECG showed biatrial enlargement and inferior ST depression. An echocardiogram showed a dilated left atrium and a mildly dilated and thickened left ventricle.

The finding of myocardial bridging was surprising. To our knowledge, this is the first report of a patient with an MYH7 mutation, RCM, and myocardial bridging. Diagnostic angiography in such patients is not routine, and the management of myocardial bridging when present in classical HCM remains controversial. Nevertheless, this case suggests that the presence of myocardial bridging should be considered with RCM and, when implicated by unexplained symptoms, should be ruled out with coronary angiography. In the absence of

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effective therapy, listing for cardiac transplantation should be considered very soon after diagnosis in this high-risk patient population.

Disclosures
None.

References


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