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 child 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 aspirin, 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 and died after an unsuccessful attempt at resuscitation.

Microscopically, there was extensive, patchy interstitial fibrosis, most prominent in the interventricular septum (Figure C) but also present in the free wall of the left ventricle. There was only minimal myocardial fiber disarray in the posterior interventricular septum. The left anterior descending was surrounded by replacement fibrosis (Figure C) with stenosed left anterior descending branch vessels because of intimal thickening (Figure D). In the interventricular septum, there were multiple foci of contraction band necrosis, some with early dystrophic calcification, indicating ongoing ischemic injury. The autopsy findings point to a restrictive, not hypertrophic, cardiomyopathy.

MYH7 mutations are most commonly associated with HCM but have been reported in RCM.1 Phenotypic and genotypic diversity is increasingly recognized in HCM and RCM, with varying degrees of hypertrophy and fibrosis perhaps representing a continuum of disease. A greater hazard for death in patients with combined RCM and HCM than for HCM alone has been described,2 and early listing for transplantation is advocated by some.3 Nevertheless, this case suggests that the presence of myocardial bridging when present in classical HCM remains controversial.4 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.5 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
effective therapy, listing for cardiac transplantation should be considered very soon after diagnosis in this high-risk patient population.

**Disclosures**

None.

**References**


**Key Words:** cardiomyopathy ■ sudden death ■ ischemia ■ pathology ■ sudden death
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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Circ Heart Fail. 2012;5:e92-e93
doi: 10.1161/CIRCHEARTFAILURE.112.969303
Circulation: Heart Failure is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 1941-3289. Online ISSN: 1941-3297

The online version of this article, along with updated information and services, is located on the World Wide Web at:
http://circheartfailure.ahajournals.org/content/5/6/e92

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