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Pathological Case of the Month

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A PREVIOUSLY healthy 6-year-old white boy was admitted to the hospital after a second episode of syncope. The first occurred several months before when while racing a sibling, he became limp and unresponsive for less than 5 minutes. On awakening, he was aware of his surroundings, but slept a great deal for the next several hours. He was fine until the day of admission when, while racing another sibling, he collapsed, became unresponsive for less than 5 minutes, and then became awake and alert. His medical history is remarkable for lack of routine immunizations, given his parents' medical convictions. There was no history of seizure disorder or cardiac arrhythmia. Family history was noncontributory.

On admission, he was a well-appearing boy. His height was 113 cm, 25th percentile; weight, 19.5 kg, 50th percentile. He had no dysmorphic features and resembled other family members. Remarkable findings included a 2/6 systolic ejection murmur maximal at the upper left sternal border that transmitted along the left sternal border as well as to the back on the left. S_1 and S_2 were normal; diastole, quiet; pulses, full and equal. Occasional premature beats were noted during auscultation. He was alert and responded to questions appropriately. Results of cranial nerve, motor, cerebellar, sensory, and deep tendon reflex tests were normal. A chest x-ray film showed a generous heart size with slight left ventricular prominence. Lung fields were clear. Electrocardiography showed sinus rhythm with frequent premature atrial contractions but no premature ventricular contractions. The QT interval was slightly prolonged at 0.45 seconds. The QRS axis was +130 with large Q waves over the right precordium and no Q waves over the left. Echocardiography (**Figure 1**) demonstrated a globular heart with left ventricular trabeculation. Shortening fraction was 28%. Hemodynamically insignificant mild pulmonary stenosis, pulmonary insufficiency, tricuspid regurgitation, and aortic insufficiency were also noted. Exercise study results were uneventful. Holter monitoring showed frequent premature atrial contractions but no evidence of supraventricular tachycardia. There were 2 premature ventricular contractions during the 24-hour monitoring. Results of an electroencephalogram were normal.

The risk of sudden death was explained to the parents. They chose to return in 1 month to pursue therapeutic options. Two weeks after discharge the patient experienced a syncopal event at home while running and playing. He collapsed and became unconscious. At the local emergency department he was apneic, cyanotic, and had weak pulses. He did not respond to full resuscitative efforts and was pronounced dead. A full autopsy was performed, which was remarkable for the 170-g (mean normal weight for age, 90 g)[1] globoid heart with deep trabeculations in both ventricular walls (**Figure 2**).

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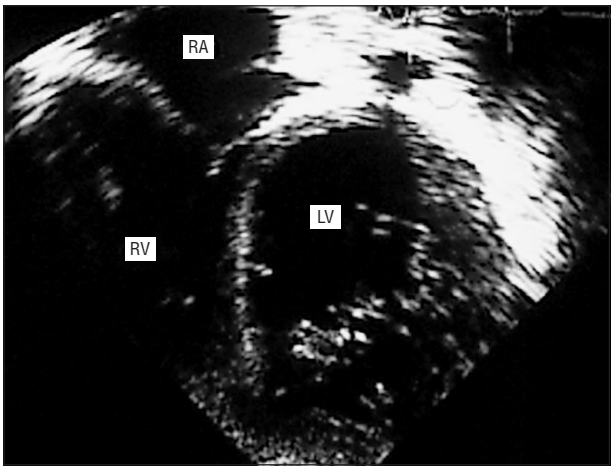

Figure 1.

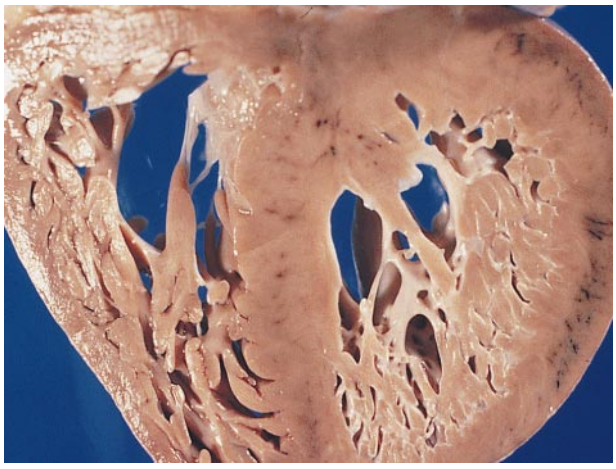

Figure 2.

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Diagnosis and Discussion

Noncompaction of the Left Ventricular Myocardium

Figure 1. *An echocardiogram in the subxiphoid view shows prominent left ventricular trabeculations with deep intertrabecular recesses involving especially the apex and free wall. RA indicates right atrium; RV, right ventricle; and LV, left ventricle.*

Figure 2. *A coronal section of the heart, with the left ventricle on the right side, shows interstices deep within the hypertrophied left ventricular free wall.*

Figure 3. *The endothelium of the left ventricular trabeculae and interstices shows fibroelastosis (hematoxylin-eosin, original magnification ×40).*

This boy had isolated noncompaction of the left ventricular myocardium (also known as persistence of spongy myocardium), a rare form of congenital cardiomyopathy in which the left ventricular wall fails to become flattened and smoother as it normally would during the first 2 months of embryonic development.[2,3] This developmental arrest results in decreased cardiac output with subsequent left ventricular hypertrophy. The aberrant left ventricular trabeculae predispose to abnormal cardiac conduction and potentially fatal cardiac arrhythmias. The interstices within the trabeculated left ventricle predispose to thrombus formation with secondary systemic embolic events. Fibroelastosis of the adjacent ventricular endocardium (**Figure 3**) is a secondary phenomenon resulting from the abnormal blood flow pattern in the left ventricular chamber. No evidence of systemic embolic events was found at autopsy; however, hemosiderin-laden macrophages in the lungs indicate evolving congestive heart failure.

The echocardiographic and computed tomographic appearance of this entity in nonlethal cases has been described.[4,5] Reported cases range from 11 months to 44 years and occur in both sexes. Associated facial dysmorphology and a variety of structural cardiac anomalies have been found in some cases.[6,7] Familial cases suggest autosomal dominant inheritance of a sporadic new mutation.

Treatment consists of anticoagulants, antiarrhythmic medications, and avoidance of strenuous exercise. Echocardiographic screening of surviving siblings is recommended.

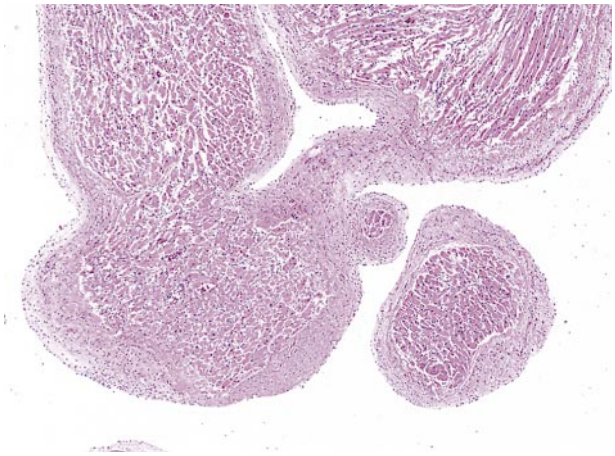

Figure 3.

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