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Brief Report

Spongy cardiomyopathy in a neonate

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Abstract Isolated noncompaction of ventricular myocardium is a rare cardiomyopathy, presumed to originate from a developmental abnormality in the evolution of the heart, and resulting in sponge-like myocardium. Isolated ventricular noncompaction can present with a variety of symptoms, but usually includes heart failure. The diagnosis is often made by echocardiography, which reveals a very distinct image of the myocardium, with many deep, confluent recesses and dense trabeculations. We encountered such findings in a moribund neonate presenting with cardiogenic shock with extremely low shortening fractions. After treatment with intravenous and oral cardiotonics, coupled with afterload reduction, we were able to optimize the balance between ventricular filling and myocardial contractility, resulting in markedly improved cardiac function as judged clinically, and as measured by echocardiography. As far as we know, this is one of the youngest patients yet reported to have a good recovery.

Keywords: Neonatal ventricular noncompaction; cardiomyopathy; crocodiles

ISOLATED NONCOMPACTION OF THE VENTRICULAR myocardium, also known as spongy myocardium, or "Mehrschwamm" cardiomyopathy in German, is a very rare cardiac malformation. Noncompaction can be isolated, or be associated with other cardiac malformations, such as obstruction of the ventricular outflow tracts, or valvar aortic stenosis or atresia. Isolated noncompaction can involve both ventricles, or only the left ventricle. As far as we know, isolated right ventricular noncompaction has never been documented. The cardiomyopathy is thought to be a result of a developmental error in the embryogenesis of the heart, with failure of compaction of loose myocardial fibres producing a sponge-like myocardium. A specific cause has not been identified, though a familial occurrence has been reported, suggesting a genetic background. Although patients may not present until old age, usually they are first seen in childhood. In this instance, we describe presentation in a neonate, and we compare the findings to the echocardiographic appearances of the ventricular myocardium in the Nile crocodile.

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Case report

A girl was born after an uneventful, full term, first pregnancy. There was no consanguinity, and no medication was used during the pregnancy. The family history of both parents was negative for cardiac disorders. The first days of life were, according to the parents, without abnormalities. She fed well, had a normal colour, and did not gain weight excessively. On the 12th day, however, she began to have problems with feeding, was irritable, pale, and cyanotic, and felt cold. She was taken to the emergency department of a regional hospital, where cardiogenic shock was recognised. The saturation of oxygen, as measured transcutaneousluy, was 50%, no pulses were palpable, and there was poor capillary refill. On further examination, there were no dysmorphic features, the heart sounds were normal, and no murmurs were audible. The liver was grossly enlarged. She was transferred to our hospital immediately.

Upon arrival, the patient remained in shock. Physical examination elicited the same findings, and analysis of blood gases revealed a combined metabolic and respiratory acidosis. A full blood count, serum glucose, and electrolytes were normal. Renal and hepatic functions were impaired due to the cardiogenic shock. Chest radiography showed an extremely enlarged heart. The electrocardiogram showed an atrial rhythm, biventricular hypertrophy, and abnormal repolarisation in all leads. Based upon these findings, the differential diagnosis was cardiogenic shock, a fulminant sepsis, or a metabolic disease.

Because of the respiratory failure, the neonate was intubated and ventilated, and the circulation was supported with intravenous dobutamine, enoximone, epinephrine and norepinephrine. Echocardiography revealed a very thick, trabeculated spongy myocardium in both ventricles, with a shortening fraction of only 6%. No other structural defects were observed (Fig. 1). Although we undertook extensive investigations to elicit the cause of this cardiomyopathy, we discovered no infectious cause. Viral and spirochetal serology was negative, as were bacteriologic cultures. Neuromuscular tests, including serum creatinine phosphokinase, electromyography, and skeletal muscular biopsy, were normal. Metabolic tests of serum and urine were within the normal range. Continuous electrocardiographic monitoring during several days did not show any arrhythmia. The heart appeared to be the only abnormal organ. There was a normally structured cerebrum and normal abdominal organs found at ultrasonography. The karyotype was normal without visible deletions.

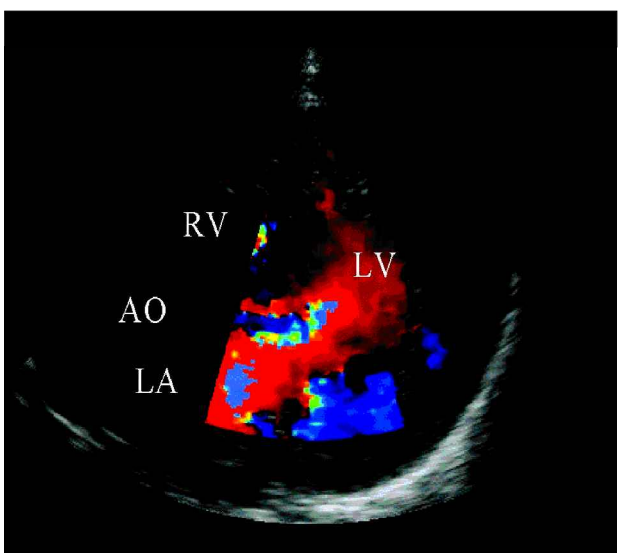

Figure 1.
Cross-sectional echocardiogram and colour-flow imaging of the left ventricle in parasternal long axis view showing the deep recesses and the spongy appearance of the thickened myocardium. LV = Left Ventricle, LA = Left Atrium, RV = Right Ventricle.

Hence, we diagnosed isolated biventricular noncompaction of the ventricular myocardium. The condition of the patient slowly improved. The intravenous inotropic support was reduced and then stopped, replacing it with oral digoxin. Diuretics and inhibitors of angiotensin converting enzyme were given to reduce the pre- and afterload. With this therapy, adequate circulation was maintained. After 9 days, the patient was extubated, and was saturating normally without supplemental oxygen. During echocardiographic follow-up, the shortening fraction had increased from 6% to 18%. Because of the risk of systemic embolus, we commenced oral acetylsalicylic acid at a dose of 10 mg daily. At present, the patient is 8 months old, and doing well. Growth and development are within the normal ranges. The prognosis in the long term, however, is uncertain and must be regarded as poor.

Discussion

Isolated noncompaction of the ventricular myocardium is a very rare malformation, first described in 1926.[1] It is believed to be a developmental derangement in the compaction of the myocardial muscle fibres in the early stages of the embryogenesis. When normal compaction fails to occur, it results in a ventricular wall with excessive trabeculations and deep recesses, giving the myocardium a spongy and hypertrophied appearance. In three-fifths of cases, the left ventricle is involved, with both ventricles affected in the remainder. Noncompaction confined to only the right ventricle has never been reported.[2] The lesion can be isolated, but is more often seen with other cardiac malformations, such as outflow obstruction, or pulmonary or aortic atresia or stenosis.[2,3]

In fish, amphibians, and reptiles, the myocardium remains spongy. Oxygenation of the myocardium occurs by direct diffusion from the ventricular cavity to the myocardial cells, or via a coronary arterial system.[4] We had the opportunity to make echocardiograms during life from the heart of a Nile crocodile weighing 150 kg. In this crocodile, the myocardial oxygen supply was studied. The animal was under general anaesthesia, and echo images were made from the epicardium. These unique images are very similar to those obtained in our patient (Fig. 2). Thus, spongy myocardium in man might be considered as a phylogenetic disorder. The cause of the developmental arrest, however, is unknown. Recent genetic research suggests a possible role for chromosome Xq28, a region in which other myopathies with cardiac involvement, such as Eimer-Dreyfuss,

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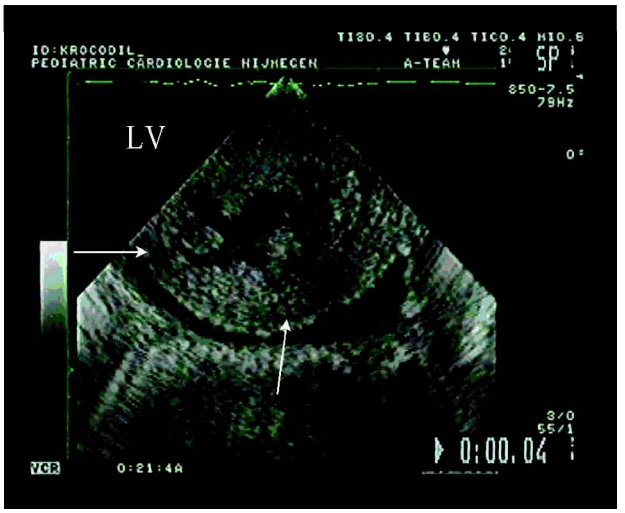

Figure 2.
Cross-sectional echocardiographic images of the left ventricle of a Nile crocodile in epicardial short axis. Note the similarities of the spongy myocardium with the echocardiographic images from our patient. LV = Left Ventricle, the arrow indicates a deep recess.

centronuclear myopathy, and Barth syndrome have all been located.[5] Recently, patients have been reported with isolated noncompaction and 5q deletion, this being the locus of the specific homeobox gene *CSX*.[6]

Familial occurrence is observed in up to half of reported cases, affecting both men and women with different family relations between them.[3,7,8] In our patient, karyotyping failed to reveal visible deletions, and the family history was negative.

The overall prevalence of isolated noncompaction is not well known. From echocardiographic studies in symptomatic patients, it is estimated to be 0.05% or less, with men being slightly more frequently affected than women.[2]

In our patient, we could not elicit any clear cause which started the events leading to the dramatic detoriation. During prolonged, continuous electrocardiographic monitoring, no arrhythmias were observed. No indication for an intervening infection was found. Probably progressive cardiac failure led to the situation encountered.

Although some patients still present in old age, the morbidity and mortality is usually substantial at an early age. Survival for five years in symptomatic patients has been as low as 50%, despite intensive monitoring and supportive therapy.[2] In some cases, therefore, transplantation may be the only curative option.[2,7]

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