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Original Article

Clinical features of isolated noncompaction of the myocardium in children

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Abstract Isolated noncompaction of the myocardium, also known as "spongy myocardium", is a rare disease in children and adults. It is suggested that, during early development of the heart, the primary spongy structure persists due to an arrest of compaction. No other cardiac malformations are found, but there are familial occurences, relations to genetic disorders or syndromes such as Melnick-needles-syndrome or Xq28-linked cardiomyopathy, and reports of conduction disorders.

We have now diagnosed isolated noncompaction in seven children aged between five weeks and 5.5 years. Three are doing well with anticongestive therapy, while transplantation of the heart was performed in one. Three of the children have died, but in only one case due to cardiac failure.

Our experience emphasises the need rapidly to establish the diagnosis, to search for associated extracardiac abnormalities, and to consider transplantation at an early stage.

Keywords: Spongy myocardium, cardiac transplantation, absence of the spleen

ISOLATED NONCOMPACTION OF THE MYOCARDIUM, or spongy myocardium, is a disorder of early cardiac development which is unassociated with other intracardiac malformations. It is characterised by prominent trabeculations and deep recesses within the compact layer of the ventricular myocardium. The recesses are lined with endothelium,[1] but have no continuity with the coronary arterial circulation. The disease is rare in children. Ichida and colleagues[2] were able to collect the data of 27 patients on the basis of sending a questionnaire to 150 hospitals in Japan with a division dealing with paediatric cardiology. Chin et al.,[1] in a single institution, were able to discover 8 cases over a period of 5 years. There are also reports in adults. Thus, from 37,555 transthoracic echocardiographic studies, 17 patients aged from 18 to 71 years were discovered with spongy myocardium. In these cases, the diagnosis had not been known before. They were referred because of unexplained heart failure, arrhythmias, and inconclusive results on previous cardiac examinations.[3] In this report, we describe our experience over five years with 7 infants and young children.

Patients and methods

Patients were screened from two regional cardiac centers over a period of 5 years, with about 9,000 echocardiographic examinations being performed each year. We detected 7 cases of spongy myocardium, and we analysed the charts of these patients retrospectively. The age at diagnosis ranged from 5 weeks to 5.5 years. There were 3 male and 4 female children. There was no evidence of occurence of any unexplained heart disease in the family histories.

We performed clinical examination along with routine electro- and echocardiography. Cardiac catheterization was undertaken in two patients, and in one of these we obtained a percutanous myocardial biopsy. Two hearts were available for direct examination, one at postmortem and the other subsequent to cardiac transplantation.

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Results

The clinical data are listed in the Table. The family histories showed no evidence of heart disease. The reason for admission to our centres was cardiac failure as the leading cause in three cases, and detection of an uncharacteristic heart murmur twice. In one patient, enlargement of the heart was noted on a chest X-ray performed because of suspected pulmonary disease, and in one the echocardiogram revealed a grossly abnormal structure of the myocardium (Fig. 1). In all patients, echocardiography showed deep recesses in the myocardium which were sufficiently marked to satisfy the criterions established by Chin et al.[1]

The clinical symptoms correlated with the severity of the observed myocardial changes. Thus, in cases with cardiac failure, there was a biventricular involvement. In asymptomatic children, suspicious findings were confined to the apical regions of the left ventricle.

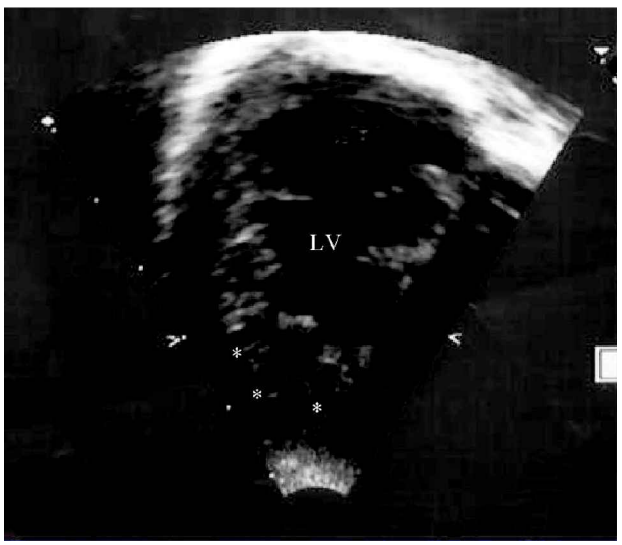

Figure 1.
Echocardiography of the left ventricle in a child, showing deep trabecular recesses at the age of 6 months.

In the patients in whom catheterisation was performed, we found pulmonary hypertension, while the angiograms confirmed the abnormal trabecular pattern of the ventricular myocardium. In one patient, we performed percutanous myocardial biopsy. This revealed deep recesses and signs of fibrosis. The examination of the heart itself, achieved in one case at autopsy and in the other subsequent to transplantation, the morphology was typical and in keeping with the clinical findings (Fig. 2). Dissection of the explanted heart revealed deep recesses in the left ventricular myocardium which were lined with endothelium.

Noncardiac problems were encountered in two cases. One 5-year-old boy suffered from severe cerebral convulsions, while one of our female patients was found to have absence of the spleen, but with no evidence of visceral heterotaxy or isomerism. Three of the children died, one because of cardiac failure, a second with complications of severe epilepsy, and because of pneumococcal infection in the third patient, who was the one with absence of the spleen. At that time, absence of the spleen was unsuspected, and antiobiotic prophylaxis was not provided.

In one boy, we undertook transplantation of the malformed heart. He, and the other three children, are currently doing well.

Discussion

Isolated noncompaction is a rare disease of the ventricular myocardium which occurs without evidence of other congenital cardiac malformations. During

Table 1.

Sex	Age at diagnosis	Symptoms	Echo	Cath.	Histology	Associated problems	Clinical course
Male	11 month	Pathолog. chest X-ray	Abnormal apical trabeculation	PHT abnorm. Trabeculation	Recesses Fibrosis		Htx
Male	5 month	Path. Echo	Abnormal LV apical trabecul.	no	no	cerebral convulsions Othahara-Syndrome	dead, cerebral reason
Female	5.5 years	Cardiac murmur	Abnormal LV apical trabecul.	no	no	no	doing well
Female	8 weeks	Cardiac failure	Abnormal LV trabecul. and function	no	postmortem	Asplenia	dead, sepsis with pneumococcus
Female	5 weeks	Cardiac failure	Abnormal LV apical trabeculation	no	no	no	doing well
Female	4 years	Cardiac failure	Abormal LV function and trabeculation	PHT abnorm. LV-trabecul.	no	no	dead
Male	3 years	Cardiac murmur	abnormal LV trabecul.	no	no	no	doing well

LV = Left ventricle; PHT = Pulmonary hypertension; Htx = Heart transplantation

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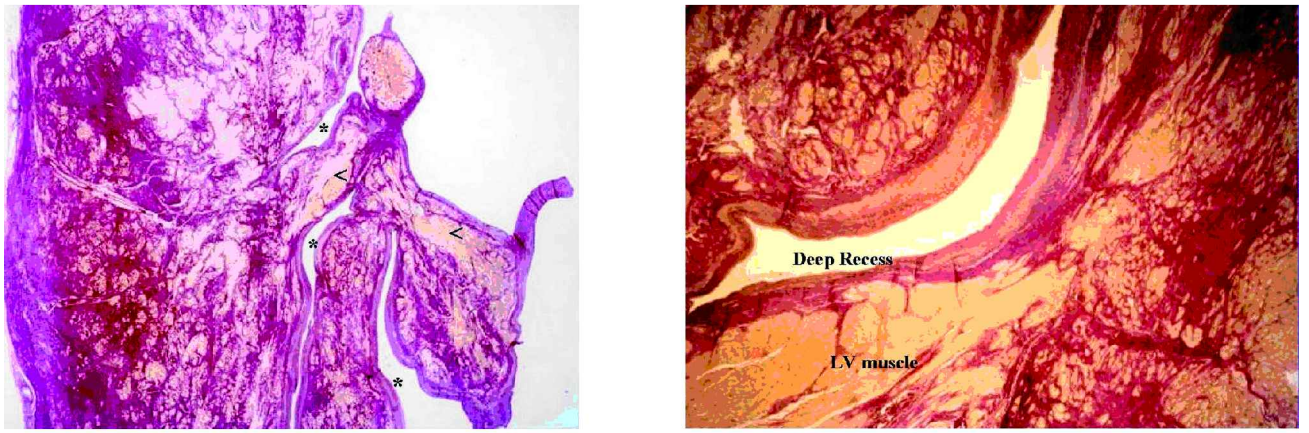

a b

Figure 2.
These histological sections, stained using the elastic – van Gieson technique, show the features of spongy myocardium. The overall view (a) shows the recesses (asterisks) extending deeply within the fibrotic myocardium (arrow heads). The higher power view (b) shows that each recess is lined with fibrous endothelium. Myocardium stains yellow, while the fibrous tissue is stained red.

early development, there is condensation of the loose mesh of musculature making up the ventricular myocardial wall. Concomitant with this, the large spaces in the trabecular meshwork flatten, almost completely in the left ventricle. It is failure of this embryonic compaction which is believed to lead to this rare myocardial disease, which produces an arrangement similar to the hearts of certain cold-blooded fishes.[4–6]

To the best of our knowledge, thus far less than 100 cases have been described in children.[7–10] The diagnosis can be established, nonetheless, using cross-sectional echocardiography alone,[1,11] this technique also revealing the extent of myocardial involvement. As yet, there is no gold standard, although diagnostic criterions have been suggested by Chin et al.,[1] and these were satisfied in our patients. It is also necessary to exclude coexisting cardiac anomalies, to visualize the prominent and excessive trabecular meshwork, and to observe the flow of blood in the intertrabecular spaces[3] in one or more of the segments of the ventricular wall.[2] Chin et al.[1] attempted to quantify these findings. Thus, they calculated a ratio for diagnosis, measuring X as the depths of the recesses in the myocardium, and Y as the thickness of the parietal wall at the peak of the trabeculations.[1] These quantitations can be carried out at the levels of the mitral valve, the papillary muscles and the apex.[2]

The problems produced by non-compaction can be due to cardiac failure,[10] thromboembolism,[5] or disturbances of rhythm.[12] Syndromic associations have been described, particularly with X-linked Melnick-Needles osteodysplasia.[13,14] Chin and his colleagues[1] also described familiar recurrences. In our patients, we observed absence of the spleen and cerebral convulsions as major non-cardiac features. The fact that the absent spleen had not previously been suspected, and the additional absence of evidence of heterotaxy, emphasises the need to focus on the possibility of extracardiac manifestations. In those cases with severe biventricular involvement and cardiac failure, transplantation of the heart should be considered at an early stage of the illness.

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