

Noncompaction with arcus aorta anomalies

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An 18-month-old girl with tetralogy of Fallot (TOF), mental retardation and multiple infarcts on brain computerized tomography (CT) is presented. Her sineangiograms showed a thrombus (moving from the left ventricle to ascending and abdominal aorta), TOF, right arcus aorta with anomalous origin of left subclavian artery from the descending aorta, honeycombed appearance of the apex and half of the posterior wall of left ventricle and the apex of right ventricle.

Her echocardiography (ECHO) examinations revealed depressed left ventricular systolic function and thrombus in the left ventricle apex, with noncompaction of the left ventricle apex and the middle portion of posterior wall, and in the right ventricular apex.

Key words: noncompaction, arcus aorta anomalies, thrombosis, children.

Noncompaction of ventricular myocardium is a rare disorder characterized by numerous excessively prominent trabeculations and deep intratrabecular recesses¹. It is usually seen in the pediatric population and is associated with other cardiac malformations. However, cases with isolated left ventricular noncompaction of left ventricular myocardium have been reported²⁻⁴.

Clinical presentations of the disease are left ventricular dysfunction (depressed systolic function or restrictive filling pattern), ventricular arrhythmia and systemic embolization²⁻⁴. Dysmorphic facial appearance and familial recurrence are also reported².

In this report, a case with tetralogy of Fallot (TOF), arcus aorta abnormalities, and motor retardation is presented. By echocardiography (ECHO) and cardiac catheterization, biventricular noncompaction and left ventricular thrombus were diagnosed.

The aim of this report is to draw attention to this rare entity.

Case Report

An 18-month-old girl with TOF was referred to our hospital for cardiac catheterization and surgery. Her family history was unremarkable. She had been followed-up for TOF and given iron therapy.

On physical examination, her length was 75 cm (25-50 p), and her weight was 7.37 kg (<3 p). Her facial features were normal. She had cyanosis, her heart rate was 135 bpm and her blood pressure was 70/40 mmHg.

Cardiac examinations revealed a thrill at the pulmonary region, increased S2, and 2/6 grade systolic ejection murmur.

The electrocardiogram (ECG) demonstrated right axis, sinus rhythm, right ventricular hypertrophy, and ST depression in the DI, DII, aVF, V5, V6 derivations.

Chest X-ray showed right ventricular prominence and decreased vascular marking. Neurologic examinations revealed motor retardation. Her brain computerized tomography (CT) examination showed multiple infarcts in the centrum semiovale and cerebellar hemispheres, and minimal cerebral atrophy.

The echocardiography showed an interventricular septal defect, overriding of aorta, infundibular and valvular pulmonary stenosis, hypoplasia of pulmonary artery, and depressed left ventricular systolic function (SF). The SF and EF of left ventricle were 26% and 60% respectively. At the cardiac catheterization, during the left ventricle angiography, a thrombus moving from the left ventricle to the ascending and the abdominal aorta

was seen. Catheterization was stopped immediately, and she was transferred to the intensive care unit. The observation revealed no neurological symptoms. Her ECHO examination was redone, and a thrombus (0.78x1.03 cm) was seen in the left ventricle apex (Fig. 1). There were excessive prominent trabeculations in the left ventricle apex and in the middle portion of the left ventricle (LV) posterior wall (Fig. 2a). The same trabecular pattern was also observed in the right ventricular apex by 2-D and color ECHO (Fig. 2b). Anticoagulant therapy (streptokinase: loading 4000 U/kg, maintenance 2000 U/kg/hr for 6 hours, heparin IV for 5 days, and warfarin 0.2 mg/kg/d PO after 5th day of heparin therapy) was started. On the 24th hour of the therapy echocardiographic study showed a decrease in the dimensions of the thrombus in the LV apex, and it disappeared on the 4th day of the therapy.

Her sineangiograms showed TOF, and right arcus aorta with anomalous origin of left subclavian artery from the descending aorta (Fig. 3). The honeycombed appearance of the inner curvature of the apex and half of the posterior wall of the LV, and the apex of right ventricle were seen on the right and the LV



Fig. 1. Thrombus in the left ventricular apex.



Fig. 2a. Noncompaction of the left ventricular apex and the left ventricular posterior wall.

angiocardiographies (Fig. 4). Beta blocking therapy was started for the infundibular stenosis. However, she died the 3rd week of therapy due to a cyanotic spell.



Fig. 2b. Noncompaction of the right ventricular apex.
*: deep trabecular recess.

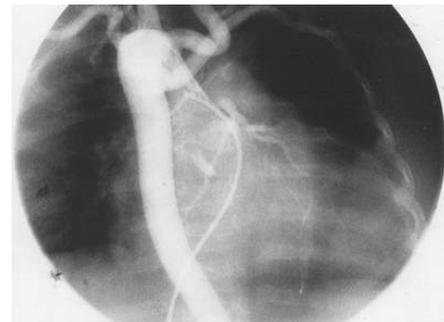


Fig. 3. Right arcus aorta with anomalous origin of left subclavian artery from the descending aorta.

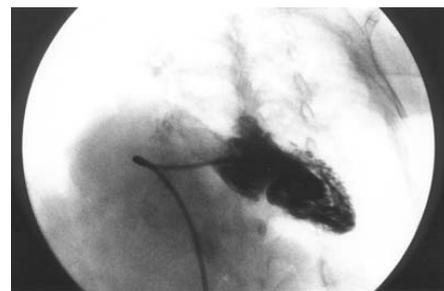


Fig. 4. Embryonic trabecular pattern of the left ventricle.

Permission for postmortem examination was not granted by the parents.

Discussion

Noncompaction of the ventricular myocardium (NVM) represents an arrest in the normal process of myocardial compaction, resulting in

the persistence of multiple prominent ventricular trabeculations and deep intertrabecular recesses⁵. Noncompaction may occur as an isolated cardiac lesion, and reports solely based on isolated noncompaction are increasing in number⁵⁻⁷. On the other hand, the persistence of noncompaction with congenital heart disease, like obstructive lesions of right or left ventricle, and anomalies of the coronary arteries, has been reported^{5,6}. Pressure overload or myocardial ischemia in the latter cases of myocardial compaction may stop the normal regression of embryogenic myocardial sinusoids. Although severe outflow tract obstruction was in the right ventricle in our case, the left ventricle suffered noncompaction. Coronary artery abnormalities were also not present. Whether the two forms of noncompaction are common or distinct lesions is not yet clear.

Both familial and sporadic forms of isolated NVM have been described⁸⁻¹⁰. The gene responsible for the X-linked familial form of isolated NVM has been localized to the X chromosome in the Xq28 region where other myopathies with cardiac involvement have been located¹¹. These myopathies (Emery-Dreifuss muscular dystrophy, myotubular myopathy, Barth syndrome) are commonly accompanied by arrhythmia⁶. Our case was the single case in her family, and genetic screening was not possible.

Nonspecific dysmorphic facial features (prominent forehead, strabismus, low-set ears, and micrognathia) are observed in some children. Our case did not have such facial features.

Clinical manifestations include heart failure due to LV systolic and diastolic dysfunctions, systemic embolization due to atrial arrhythmias, intraventricular thrombi located within deep intertrabecular recesses, and ventricular arrhythmias²⁻⁶.

This case had LV systolic dysfunction and intraventricular thrombus. It is probable that the case had central nervous system involvement due to systemic cardioembolic event. Since the diagnosis of the thrombus by ECHO may be difficult due to multiple trabeculations, prophylactic anticoagulant therapy should be encouraged for the prevention of the embolic episodes. Also echocardiographic screening must be carried out in the first-degree relatives to identify noncompaction in the asymptomatic phase.

Left ventricular (LV) dysfunction in this entity may be due, to systolic or diastolic dysfunction^{2,3-5}. Therefore special care should be taken regarding the differential diagnosis of young patients with restrictive cardiomyopathy³.

Robida and Hajar⁴ reported two children with severe heart failure and left bundle branch block. They advised that noncompaction should be added to the list of the causes for the left bundle branch block in children. According to the study of Ichida et al.⁶, a higher incidence of Wolff-Parkinson-White (WPW) syndrome was seen in children with noncompaction, whereas left bundle branch block was rarer in children than in adults with noncompaction.

Clinical manifestations like myocardial failure, arrhythmia, and thromboembolism, which represent a detailed differential diagnosis, determine the outcome. Early diagnosis of patients may be lifesaving: anticoagulant therapy for patients should be encouraged and screening of first-degree relatives should be carried out.

REFERENCES

- Jenni R, Goebel N, Tartini R. Persisting myocardial sinusoids of both ventricles as an isolated anomaly: echocardiographic, angiographic, and pathologic anatomical findings. *Cardiovasc Intervent Radiol* 1986; 9: 127-131.
- Chin TK, Perloff JK, Williams RG, Jue K, Mohrmann R. Isolated noncompaction of left ventricular myocardium, a study of eight cases. *Circulation* 1990; 82: 507-513.
- Hook S, Ratliff NB, Rosenkranz E, Sterba R. Isolated noncompaction of the ventricular myocardium. *Pediatr Cardiol* 1996; 17: 43-45.
- Robida A, Hajar HA. Ventricular conduction defect in isolated noncompaction of the ventricular myocardium. *Pediatr Cardiol* 1996; 17: 189-191.
- Aagmon Y, Connolly HM, Olson LJ, Khandheria BK, Seward JB. Noncompaction of the ventricular myocardium. *J Am Soc Echocardiogr* 1999; 12: 859-863.
- Ichida F, Hamamichi Y, Miyawaki T, et al. Clinical features of isolated noncompaction of the ventricular myocardium: long-term clinical course, hemodynamic properties, and genetic background. *J Am Coll Cardiol* 1999; 34: 233-240.
- Matsuda M, Tsukahara M, Kondoh O, Mito H. Familial isolated noncompaction of the ventricular myocardium. *J Hum Genet* 1999; 44: 126-128.
- Kurosaki K, Ikeda U, Hojo Y, Fujikawa H, Katsuki T, Shimada K. Familial isolated noncompaction of the left ventricular myocardium. *Cardiology* 1999; 91: 69-72.

9. Pauli RM, Scheib-Wixted S, Cripe L, Izumo S, Sekhon GS. Ventricular noncompaction and distal chromosome 5q deletion. *Am J Med Genet* 1999; 85: 419-423.
10. Rolf E, Bender F. Identification of a rare congenital anomaly of the myocardium by two dimensional echocardiography: persistence of isolated myocardial sinusoids. *Am J Cardiol* 1984; 53: 1733.
11. Bleyl SB, Mumford BR, Brown-Harrison MC, et al. Xq28-linked noncompaction of the left ventricular myocardium. Prenatal diagnosis and pathologic analysis of affected individuals. *Am J Med Genet* 1997; 72: 257-265.