Congenital Diverticulum of the Left Ventricle Associated with Hypoplastic Right Ventricle and Ventricular Septal Defect

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SUMMARY
A case of congenital diverticulum of the left ventricle associated with a ventricular septal defect and severe hypoplasia of the right ventricle without tricuspid or pulmonary atresia is described and the relevant literature is reviewed. The distinction between true muscular diverticulae and congenital apical aneurysms is made, the latter term being preferred for similar abnormalities described as fibrous diverticulae or congenital epicardial cysts. Rupture is rare in true muscular diverticulae and the high pressure recorded in our case is thought to have been caused by catheter entrapment.

Additional Indexing Words:
Ventricular aneurysm
Fibrous diverticulae
Pulmonary atresia
Valvular atresia
Epicardial cysts

CONGENITAL DIVERTICULUM of the left ventricle is a rare anomaly usually accompanied by other cardiac and extracardiac malformations.

Isolated hypoplasia of the right ventricle unassociated with tricuspid or pulmonary valve atresia is a well-delineated congenital cardiac malformation which is frequently associated with an atrial or a ventricular septal defect and has been described in siblings.1-5

This paper reports the combination of isolated hypoplasia of the right ventricle, ventricular septal defect, and a diverticulum of the left ventricle, and reviews the literature pertaining to cardiac diverticulae.

Case Report

A two-month-old Negro girl was referred for investigation of a pulsatile umbilical tumor which had been present since birth. She was the first child of normal parents and there was no history of heart disease in the family.

Examination revealed a well-looking infant with no evidence of cardiac failure. There was no clinical cardiomegaly, and no thrills were palpable. On auscultation a short grade 2/6 ejection systolic murmur was audible at the left parasternal border.

A markedly pulsatile mass was present at the umbilicus which on palpation was found to protrude through an umbilical hernia. It felt elongated, saccular, and extended proximally toward the diaphragm. Compression of its proximal portion failed to reduce distal pulsation. No murmur was audible over the mass. There was associated diaphragmation of the rectus abdominis.

A roentgenogram of the chest showed a right-sided cardiac apex with the right hemidiaphragm lower than the left. There was no cardiomegaly and the lung vascularity was within normal limits. A large thymic shadow was present. The visceral situs was normal (fig. 1). The electrocardiogram showed sinus rhythm with a normal P wave vector, a mean frontal plane QRS axis of +220°, and biventricular hypertrophy.

The clinical diagnosis was that of a congenital diverticulum of the left ventricle probably associated with a ventricular septal defect.

Cardiac catheterization demonstrated equal ventricular pressures (79/8–12 mm Hg) The aortic pressure was 75/38 mm Hg and the pulmonary artery pressure 77/30 mm Hg. A diverticulum was entered at the apex of the left ventricle; the pressure recorded in the diverticulum was 86/6–14 mm (fig. 2). The configuration of the pressure pulse in the diverticulum was suggestive of catheter entrapment. Left ventricular cineangiography and injection of contrast medium into
the diverticulum demonstrated its continuity with the apex of the left ventricle and also the presence of a ventricular septal defect and a hypoplastic right ventricle (fig. 3).

During hospitalization the child died because of bilateral bronchopneumonia unresponsive to antibiotic and oxygen therapy.

Pathologic Findings

The lungs showed the presence of bilateral bronchopneumonia.

The sternum was short and the xiphisternum was rudimentary. A diverticulum arose as a tubular extension of the apex of the left ventricle and consisted of all layers of the wall of the left ventricle with a smooth endocardial lining. The diverticulum protruded through a defect in the diaphragm into an umbilical hernia and was attached to the subcutaneous tissue in this area. The right ventricle was severely hypoplastic with virtual absence of the inflow portion; the tricuspid valve was also markedly hypoplastic. The pulmonary valve was normal. An infracristal ventricular septal defect was present (fig. 4).

Discussion

Congenital diverticulum of the heart consists of an out-pouching of either ventricle including...
CONGENITAL DIVERTICULUM OF LV

Figure 3

Cineangiogram, anteroposterior projection. Dye injected into the diverticulum (D) outlines left ventricle (LV) and hypoplastic right ventricle (RV) via a ventricular septal defect (between arrows.) The left atrium (LA) is also opacified.

endocardium, myocardium, and occasionally, pericardium. Diverticulae may arise from the left,6-10 more rarely the right,11, 12 or even from both ventricles.13 Associated intracardiac malformations and defects in the abdominal wall are common. They are thus different from "fibrous diverticulae" and "congenital epicardial cysts," which may occur in the apical as well as the subvalvular positions of the left ventricle. The latter appear to be the result of a congenital defect which forms the basis for subsequent formation of fibrous false aneurysms which are unassociated with other cardiac defects and have a tendency to rupture.14-20 We would prefer to call the latter "congenital apical or subvalvular aneurysms" to distinguish them from muscular diverticulae of the type reported here.

A diverticulum arising solely from the right ventricle is rare. Two cases described by Carter12 appear to be true diverticulae of the right ventricle but differ from the left ventricular type in that they were large, and had wide ostia situated in the lateral wall of the right ventricle. The clinical presentation in each patient suggested tetralogy of Fallot and the diverticulae were only detected by angiography. Pathologically, both cases showed diverticulae containing endo- and myocardium. The case reported by Cumming11 was different in that the ostium was situated at the apex of the right ventricle and the wall of the diverticulum consisted of endocardium and a thick layer of collagen tissue. We would prefer to call this an apical aneurysm of the right ventricle similar to those described in the left ventricle.14-16

A congenital diverticulum arising from both ventricles has also been described. The diverticulum penetrated the diaphragm and presented in the epigastrium; this was associated with a thin slit-like rudimentary right ventricle, tricuspid atresia, pulmonary artery hypoplasia, and a ventricular septal defect.13

Congenital diverticulum of the left ventricle is less rare. The clinical presentation is usually typical and the diagnosis may be made at the bedside.1, 2 The diverticulum usually presents in the epigastrium or at the umbilicus as a pulsatile tumor, which when compressed, continues to pulsate in the proximal and distal positions, emphasizing the myocardial content of the tumor. Associated cardiac and extracardiac anomalies have been reported. A ventricular septal defect is almost invariably present. Other abnormalities reported include tricuspid atresia,21, 22 patent ductus arteriosus, atrial septal defect, and truncus arteriosus with pulmonary stenosis.23

A syndrome frequently associated with diverticulum of the left ventricle has been described by Cantrell24 and others25, 26 and consists of

(1) malrotation of the heart with dextroposition;
(2) intracardiac anomalies including ventricular septal defect and diverticulum of the left ventricle;
(3) pericardial defect;
(4) diaphragmatic defect;
(5) deficient abdominal wall with diastasis of the rectus abdominis and an umbilical hernia;
(6) defective lower sternum.

Most of these features were present in our case but the additional intracardiac anomaly was a hypoplastic right ventricle.

Congenital hypoplasia of the right ventricle is usually associated with tricuspid atresia,24 pulmonary atresia with intact ventricular septum,25 or both tricuspid and pulmonary valve atresia. Isolated hypoplasia of the right ventricle unassociated with the aforementioned valvular abnormalities is
uncommon but has been described in association with atrial or ventricular septal defects and may occur in siblings.\textsuperscript{3-7} Our case represents the unreported combination of isolated hypoplasia of the right ventricle, ventricular septal defect, and a diverticulum of the left ventricle.

The embryological derivation of cardiac diverticulae is uncertain. Carter et al.\textsuperscript{12} postulated that right ventricular diverticulae of the form they described may represent an abnormal out-pouching of the primitive ventricle comparable to that occurring during the development of the normal right ventricle. The muscular diverticulae arising from the apex of the left or right ventricle probably have a different origin as described by Parsons.\textsuperscript{13}

These may result from anomalous development at about the fourth week of embryonic life when the heart wall may become attached through the septum transversum to the structures forming the yolk sac and then becomes drawn out as the embryo rapidly grows and unfolds. This may account for the defects in the diaphragm, sternum, and anterior abdominal musculature. Explanations incriminating weakness of the cardiac apex\textsuperscript{17} appear to be applicable to congenital apical aneurysms and cannot account for the syndrome complex such as described here and elsewhere.

Surgical treatment has generally been advised in view of the danger of rupture or the occurrence of fatal arrhythmias. The danger of rupture may have

Figure 4

Specimen of heart (A) showing diverticulum (D) which arises as a tubular extension of the apex of the left ventricle (LV). The right ventricle (RV) has a grossly thickened wall but its inflow portion is markedly hypoplastic. (B) View of left ventricular cavity showing ventricular septal defect (between arrows). LA = left atrium; Ao = aorta; PA = pulmonary artery.
been overemphasized by the inclusion of cases of congenital apical aneurysms, i.e., fibrous diverticulae or congenital epicardial cysts.\textsuperscript{11-20} Lowe has postulated that the higher pressure reached in the diverticulum (in his case measured by direct puncture) may account for rupture.\textsuperscript{8} We feel that the higher pressure recorded in our case may, however, be a form of "catheter entrapment." The cosmetic improvement and danger of direct trauma alone, however, appear to be sufficient indication for resection of the diverticulum and repair of the extracardiac defects. Associated cardiac anomalies require assessment and treatment on their own merit. Surgical excision of the diverticulum had been planned in our case but the severe hypoplasia of the right ventricle would have made total correction impossible.

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