Congenital diverticulum of the left ventricle

Report of two unusual cases

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SUMMARY Of two infants with congenital cardiac diverticula one had a defect consisting of a sub-mitral valvular diverticulum rather than the expected aneurysm. It was clearly shown by echocardiography as well as angiography and was associated with moderate mitral incompetence and a poorly functioning left ventricular wall. The second infant had an isolated apical left ventricular diverticulum with subnormal left ventricular function and severe mitral incompetence. Because of intractable congestive heart failure this infant underwent successful replacement of a dysplastic fibrotic mitral valve which on histological examination had myxomatous features. The association between congenital mitral disease and congenital apical diverticula may be related to race and sex.

Congenital diverticulum of the heart is a rare condition, with a prevalence of 0-4% in 750 necropsies of cardiac cases. The diverticulum is perhaps produced by an outpouching of endomyocardium in the first two or three weeks of embryonic life at a weak region of the right ventricular or left ventricular wall or in the muscular septum. Apical diverticulum of the left ventricle has been reported in 82 patients; most of them had a mid-line thoracoabdominal lesion and congenital heart disease. Only 25 of the 82 patients had an isolated apical defect. Subvalvular fibrous wall aneurysm is another infrequent form of left ventricular defect seen mostly in black Africans. I report two unusual cases of cardiac diverticula, one with a sub-mitral lesion which fulfils the criteria for a diverticulum rather than the expected aneurysm, the other with an apical diverticulum without the midline defects and with unexpected mitral valve pathology.

Case reports

CASE 1
A 1½ year old Saudi girl with a harelip and cleft palate had recurrent episodes of dyspnoea, feeding difficulties, and slow weight gain since early infancy. Her mother had had a viral illness for two weeks during the first six weeks of pregnancy. On cardiac examination of the child, the point of maximum impulse was felt at the fifth left intercostal space with a slight increase in the left ventricular impulse. Grade II/VI pansystolic and grade I/VI mid-diastolic murmurs were heard at the apex and extended towards the left axilla. The electrocardiogram showed left ventricular hypertrophy, a deep Q wave with inverted T waves in leads II, III, and aVF, and slight ST segment elevation in the anterior and depression in the left precordial leads (Fig. 1). Chest x ray films showed generalised cardiomegaly with a mild degree of pulmonary venous congestion. Echocardiography confirmed the electrocardiographic findings and showed decreased left ventricular function with fractional circumferential fibre shortening of about 20%. A sweep from the apex to the outflow tract of the left ventricle showed posterior displacement with thinning of the left ventricular posterior wall starting at the sub-mitral valve area and an infero-posterior extension towards the apex, suggesting an outpouching of the posterior wall (Fig. 2 a and b). Right and left heart catheterisation showed a pulmonary pressure (systolic/diastolic/mean) of 34/14/22 mm Hg, an aortic pressure of 84/45/61 mm Hg, and a left ventricular pressure of 81/11 mm Hg (systolic/diastolic/end diastolic); oxygen pressure was 101 mm Hg and cardiac index 3.4 l/min/m². Cineangiography showed a notably dilated and poorly functioning left ventricle with an ejection fraction around 30% and a mild to moderate degree of mitral regurgitation. A large multilobulated "diver-
A diverticulum* with a trabeculated muscular wall was seen arising from the inferolateral aspect of the left ventricle at the junction between the left ventricular posterior wall and the interventricular septum, starting below the mitral valve ring and extending inferoposteriorly (Fig. 3a and b). The wall of the diverticulum was contractile with slight paradoxical motion at end systole. The aortic angiogram showed a normal origin and distribution of the right and left coronary arteries (Fig. 4). The circumflex arteries, however, were extended and stretched around the margin of the left ventricular diverticulum (arrow). The child was discharged taking digoxin. During a two year follow up, she gained some weight and underwent successful surgical repair of the harelip and cleft palate. The clinical symptoms and the size of the heart and diver-
Congenital diverticulum of the left ventricle

Fig. 3 Left ventricular angiogram (case 1) in (a) the left anterior oblique projection in systole (LVs) showing the large diverticulum (D) extending inferoposteriorly from the submtral valve area; the diverticular wall (arrows) appears multilobulated with dye free filling defects representing either clot or trabeculae; and (b) the right anterior oblique projection in systole (LVs) clearly showing a large cauliflower shaped diverticulum (D) with an inferoposterior extension and multilobulated wall. Note a large left ventricular cavity with decreased function.

ticulum, however, remained unchanged as seen on chest x ray films and echocardiogram.

Fig. 4 Aortic root angiogram (case 1) in the left anterior projection showing pronounced stretching and posterior displacement of the circumflex branch of the coronary artery.

CASE 2

A 1 year old Saudi girl was referred to this hospital for cardiac evaluation because of failure to thrive, recurrent respiratory infection, circumoral cyanosis, and dyspnoea at rest. Her mother reported “not feeling healthy” during the first trimester of pregnancy with generalised weakness, frequent episodes of abdominal pain, and fatigue. At birth the child had a short period of cyanosis that resolved spontaneously. On admission she was small for her age, emaciated, and disorientated. Her skin colour was ashen, and there was a moderate degree of respiratory distress. The point of maximum impulse was at the sixth to seventh left intercostal space, 2 cm outside the mid-clavicular line. A grade III/VI pansystolic murmur was best heard at the apex with radiation to the left axilla. The electrocardiogram showed a deep Q wave in leads II, III, and aVL and the left precordial leads. Chest x ray films showed a pronounced generalised cardiomegaly with a cardiothoracic ratio of 70%. The pulmonary veins appeared congested with atelectasis of the left lower lobe. After initial treatment with digoxin and frusemide, she underwent cardiac catheterisation, which showed normal pressures in the right heart. The pressures in the left ventricle (systolic/diastolic/end diastolic) were 93/0/9 mm Hg and in the aorta (systolic/diastolic/mean) 97/53/72 mm Hg; and the pulmonary capillary wedge pressures were A=16 and
Fig. 5  Left ventricular cineangiogram (case 2) in right anterior oblique view showing apical diverticulum delineated by the two arrows. The mitral valve appears thickened, deformed, and incompetent. LA, left atrium; LV, left ventricle.

v = 15 (mean 9 mm Hg); cardiac index was 3.3 l/min/m²; mitral area index was 1.59 cm²/m²; oxygen pressure 77 mm Hg with an oxygen saturation of 94%.

A left ventricular cineangiogram in the right anterior view (Fig. 5) showed dilated left ventricular and left atrial cavities, a severe degree of mitral regurgitation, and decreased left ventricular function, with normal origin and distribution of the coronary arteries. A large apical left ventricular diverticulum was seen contracting normally except for a slight paradoxical motion at end systole. The infant underwent open heart surgery for mitral valve replacement with a 25 mm Björk-Shiley concave prosthesis. The diverticular wall was hard and contracting well, and so the diverticulum was not excised. The mitral valve was dysplastic and fibrotic with a poorly formed subvalvular mechanism. Microscopical examination of the valve showed extensive mucoid changes with myxomatous formation. The ventricular muscle showed focal vacuolar degeneration.

Discussion

In the past the terms congenital aneurysm and diverticulum of the left ventricle were used synonymously. Treistman et al classified the defect as a diverticulum when its root of connection to the left ventricle was narrow and was associated with congenital mid-line lesions and as an aneurysm if the connection was wide and there were no other congenital defects. In 1974 Hoefl reviewed extensively the cases of left ventricular diverticula with the aim of reclassification. Nevertheless, the generally accepted definition of a diverticulum is that its wall contains three cardiac layers (endocardium, myocardium, and pericardium) and contracts normally. It may be regarded as congenital when an apparent acquired aetiology is not evident, when the diverticulum is associated with other mid-line lesions, or when papillary muscle is involved in a contractile muscular diverticulum. On the other hand, an aneurysm consists of a fibrous sacculated lesion with paradoxical contraction of its wall. An aneurysm may be termed "acquired" when it develops as a result of infection, ischaemia, trauma, postoperative coronary disease, or other such factors. Furthermore, a congenital aneurysm is usually associated with a congenital defect or absence of muscle or an anomalous left coronary artery arising from the pulmonary artery. A false aneurysm, however, is produced by trauma, tuberculosis, or ruptured abscess and is walled by pericardium. In 1976 Gueron et al proposed a new classification of left ventricular aneurysm and diverticulum based on angiographic or postmortem examination and the location and histological characteristics of the defects. These workers reported the first two asymptomatic cases of left ventricular apical diverticulum occurring in association with mitral incompetence found in two Bedouin girls from the Middle East. They classified their two patients in the diverticular group of "unknown pathology". The pathology in the second patient in the present study is identical to that of Gueron's two patients. Furthermore, the mitral valve in this patient appeared dysplastic and fibrotic with extensive mucoid and myxomatous formation. The diverticular muscle biopsy showed focal vacuolar degeneration. This report is, therefore, the first to describe the pathological findings in this condition, and the first to prove, by surgical and histological examination, that mitral valve regurgitation is secondary to an organic disease of the mitral valve apparatus rather than the result of papillary muscle involvement in the diverticulum. Furthermore, the patient in this report was severely symptomatic, in contrast to Gueron's two asymptomatic patients, even though all three patients came from the same semitic ethnic group in two neighbouring countries in the Middle East. These observations may suggest that this type of lesion is more prevalent or even restricted to the female semitic ethnic group and is similar to the subvalvular fibrous aneurysm which is seen mainly in people of African origin. The partially contractile left ventricular diverticulum with a thick wall in case 1 differs from the fibrous aneurysm seen in black Africans but is identical to the findings in the
two infants first described by Hemsath et al\textsuperscript{16} in 1936 and by Franco-Vasquez \textit{et al}\textsuperscript{17} in 1970. The present patient also had a harelip and cleft palate and a poorly functioning left ventricle. This combination of anomalies, which has never been reported previously, may indicate an insult to the foregut and the mesodermal cell transformation to myocardium in the first three weeks of the embryonic phase. This insult may be viral in origin since it has been reported in rubella myocarditis and congenital cardiomyopathy.\textsuperscript{2 5 8 12 13 15 18 19}

It creates a structurally weak point during the development of the ventricular wall, thus serving as a base for the gradual expansion of the diverticulum with the continual increase in intracardiac pressure.\textsuperscript{8 12 15} This mechanism has been suggested by Franco-Vasquez \textit{et al}\textsuperscript{17} in the subvalvular form, and by Potts \textit{et al}\textsuperscript{20} and Swyer \textit{et al}\textsuperscript{21} in the apical form. Paronetto and Strauss\textsuperscript{13} reported a rare association of a localised left ventricular aneurysm in an 8 month old infant, resulting from a congenital muscle defect and fibroelastosis. Hemsath and others assumed that the aetiology of the diverticulum was due to excessive primordial cell stimulation at a certain point in an attempt to produce a double heart which ended in producing one or more small or large diverticula.\textsuperscript{6 11 16}

Most patients with an isolated left ventricular diverticulum have mild or no symptoms and require no surgery. Only a few patients with this condition who have symptoms of intractable congestive heart failure, life threatening arrhythmias, severe mitral incompetence, or a large akinetic aneurysm causing thrombosis or diminished left ventricular output have required surgery. The clinical presentation in the first patient in this study was a mild degree of congestive heart failure that was controlled with maintenance digoxin therapy. The second patient, however, presented with intractable congestive heart failure that required mitral valve replacement; the apical diverticulum, however, was not excised surgically because it was small in size and contracting well. The electrocardiographic changes in both patients were similar to those described in patients with left ventricular diverticula.\textsuperscript{8 14} These changes may not be of any help in small diverticula and of little help in making the diagnosis even in the presence of an extensive diverticulum.\textsuperscript{6 14} M mode echocardiography could be helpful in making the diagnosis,\textsuperscript{22} although a false negative result is a possibility. Cross sectional echocardiography may, however, be of more help, especially in the diagnosis of an interventricular septum, left ventricular wall, or subvalvular aneurysm or diverticulum.\textsuperscript{23} Roentgenograms of the chest may show only cardiomegaly or protrusion at the region of the aneurysm, which could be a clue to the diagnosis.\textsuperscript{4 10 14} Patients with small diverticula may have a totally normal chest radiograph.\textsuperscript{6} Cardiac catheterisation and angiography appear to be the only reliable and conclusive procedures for diagnosing the disease and its associated cardiac lesions and for determining the need for surgery.

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