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Brief Report

Cardiac diverticulum and omphalocele: Cantrell's pentalogy or syndrome

Feico J. J. Halbertsma,[1] Anton van Oort,[2] Frans van der Staak[3]

[1]*Department of Paediatrics,* [2]*Children's Heart Centre,* [3]*Department of Paediatric Surgery, University Medical Centre St Radboud, Nijmegen, Netherlands*

Abstract Omphaloceles and left ventricular diverticulums are rare disorders. Although either is known to occur on its own, the combination is highly suggestive of the so-called pentalogy of Cantrell. This syndrome is a combination of deformities involving midline structures, with exteriorisation of the heart, or 'ectopia cordis', as the most severe malformation. A cause has yet to be identified, though genes located on the X-chromosome may be involved. We discuss a neonate who presented with an omphalocele and a palpable diverticulum of the left ventricle. An omphalocele, especially when above the umbilicus, is an indication for further investigation for deformities as seen in the spectrum of Cantrell's pentalogy, especially cardiac malformations and anterior diaphragmatic herniation. A left ventricular diverticulum is usually associated with Cantrell's syndrome. When found, it is usually accompanied by other intracardiac malformations, so that again further examination is indicated. In our patient, there was an atrial septal defect within the oval fossa, along with a ventricular septal defect and unobstructed albeit anomalous venous pulmonary return to the left atrium. Early surgical intervention seems to be indicated, as spontaneous rupture, arrhythmias, and thromobogenicity of the ventricular diverticulum have all been reported.

Keywords: Pentalogy of Cantrell; cardiac diverticulum; omphalocele

Defects of the abdominal wall, such as an omphalocele, are frequently associated with cardiac malformations. In 1958, Cantrell described 21 patients with five abnormalities: a supra-umbilical defect of the abdominal wall in the midline, an anterior diaphragmatic hernia, sternal deformities, pericardial defects, and cardiac malformations.[1] This combination is now known as the pentalogy of Cantrell, and represents a combination of malformations in the spectrum of thoraco-abdominal midline defects. We discuss in this report a patient with a diverticulum of the left ventricle, palpable through an omphalocele, and an anterior diaphragmatic hernia. Although both lesions exist in isolation, this combination should always raise the suspicion of Cantrell's syndrome. As anterior diaphragmatic hernia is present in over half the patients having the syndrome, and intracardiac malformations in over two-thirds, thorough investigation for these anomalies is indicated.[2,3] In our patient, we found an interatrial communication, a ventricular septal defect, and elongated but unobstructed venous pulmonary return to the left atrium. Although there is no consensus in the literature about optimal treatment of the diverticulum, we chose to excise it combined with surgical correction of the omphalocele.

Case report

Our patient, a male, was the first born after a full term pregnancy without complications. The family history of the mother was uneventful, no medication having been used. At birth, a large supra-umbilical omphalocele, with cranially displaced umbilical vessels, was found. It was covered with a thin layer of skin, through which a pulsating, vascular structure was felt (Fig. 1). Upon examination, a systolic murmur, grade 2 out of 6, was heard at the apex of the

Correspondence to: Anton van Oort MD, Children's Heart Centre, University Medical Centre St Radboud, Post Box 9101, 6500 HB Nijmegen, Netherlands. Tel: (0)24 36190 60; Fax: (0)24 36190 52; E-mail: A.vanOort@ckskhc.azn.nl

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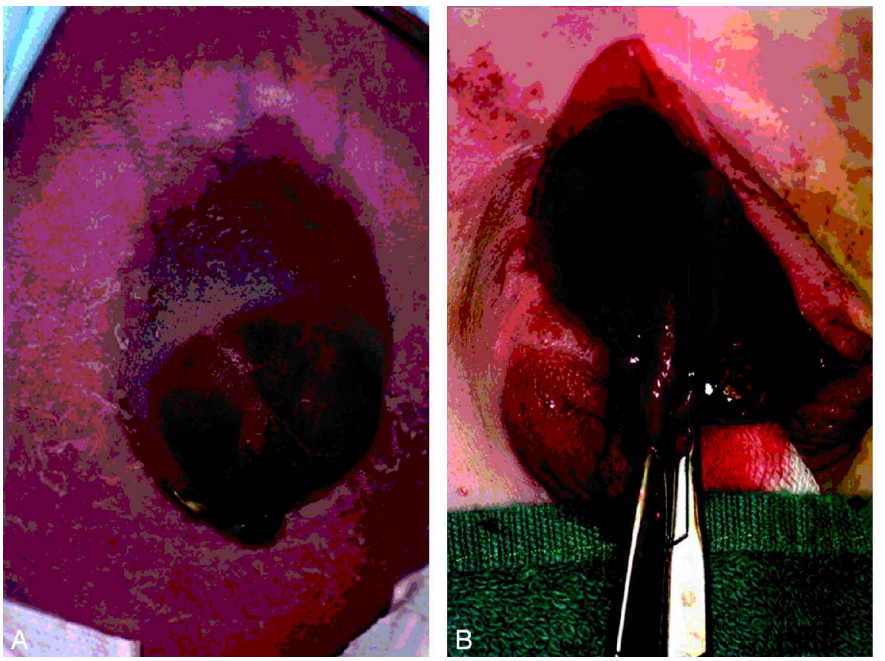

Figure 1.
(A) A large omphalocele is seen, covered by a thin layer of skin. A pulsating left ventricular diverticulum was palpable through the skin. (B) Peroperative photo showing the opened omphalocele, exposing the liver and the left ventricular diverticulum.

heart and the pulsating structure. The circulation and oxygenation were normal. Apart from these findings the examination was normal.

Chest radiography showed a slightly enlarged heart, while ultrasound revealed an anterior herniation. The electrocardiogram had increased voltages in the right precordial leads, suggesting right-sided cardiac position. No disturbances of rhythm or conduction were present. Echocardiography showed the heart to be positioned in the middle of the thorax, with the apex deviated to the right. Atrioventricular and ventriculo-arterial connections were concordant. There was an atrial septal defect within the oval fossa, and the pulmonary veins were elongated, but draining in unrestrictive fashion to the left atrium. A large, narrow based, vascular structure with bi-directional flow was seen to originate from the left ventricular apex. No ventricular septal defect was visualised. Catheterisation revealed a left ventricular apical diverticulum, a perimembranous ventricular septal defect, an atrial septal defect within the oval fossa, and equal left and right ventricular pressures (Fig. 2).

Having opted for surgical correction, the surgeon discovered an anterior diaphragmatic hernia, a pericardial defect, and a left ventricular diverticulum. The diverticulum was resected, the pericardial and diaphragm defects were covered with peritoneum, and the abdominal wall defect was closed with a Gore-Tex patch. The postoperative course was uneventful. During follow-up at the age of 7 months, the patient was doing well. A rough systolic murmur was still present, emanating from the ventricular septal defect, which was confirmed with echocardiography.

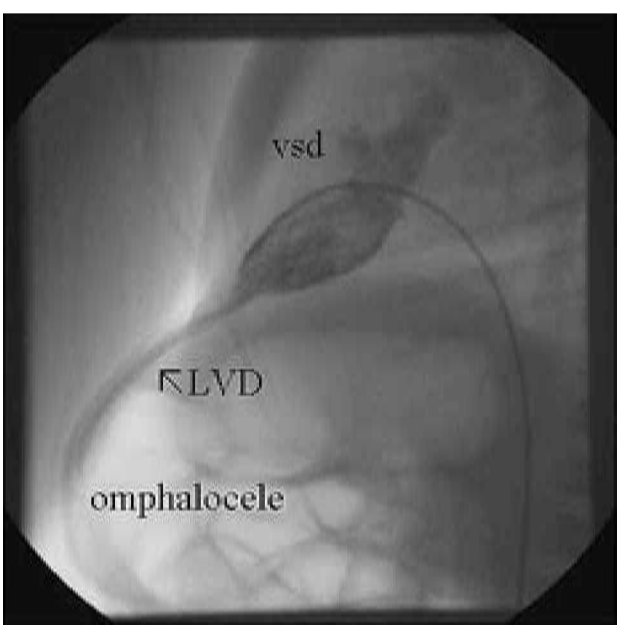

Figure 2.
Angiography, showing the left ventricle with a large diverticulum originating from its apex and extending into the omphalocele. There is a left-to-right shunt through the ventricular septal defect.

The interatrial communication and the elongated pulmonary veins also persisted.

Discussion

Omphaloceles and left ventricular diverticulums are rare disorders. Such diverticulums may occur in

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Table 1.

Syndromes with omphalocele

Cantrell's pentalogy
Beckwith-Wiedemann
Shprintzen-Goldberg
Thoraco abdominal syndrome
Omphalocele-exstrophy-imperforate anus-spinal defects complex
Melnick-Needles osteodysplasty
Cranioorodigital syndrome
Meckel syndrome type 1
Miller-Dieker lissencephaly syndrome

isolation, in which case they are usually detected in childhood, although cases have been reported in neonates and adults, with some patients even remaining asymptomatic.[4,5] More often, the diverticulums are associated with other malformations, in which case they are typically syndromic. The same applies to omphaloceles (Table 1).

Cantrell's pentalogy, the most frequent syndrome involving left ventricular diverticulums and supra-umbilical midline defects of the abdominal wall, is currently considered to be a spectrum of thoraco-abdominal midline defects, with defects of the abdominal wall at one end and exteriorisation of the heart, or "ectopia cordis", at the other.[6] Malformations can be diverse. A bifid or partially aplastic sternum is seen in three-fifths, various defects of the abdominal wall in four-fifths, and diaphragmatic and cardiac defects in just over half. Pericardial defects occur in just over two-fifths of cases.[2] Whether these malformations in various combinations are considered incomplete manifestations of the pentalogy, or called Cantrell's syndrome, is a semantic issue, but all are in the spectrum of thoraco-abdominal defects.

The cause of the midline defects has yet to be identified. A derangement in the development of the transverse septum during the third week of development probably results in a defect of the diaphragm and pericardium, and a derangement of the splanchnic mesoderm produces the cardiac malformations. Incomplete regression of the physiologic umbilical herniation of the intestines produces the omphalocele.[2] There are indications that genes located on the X-chromosome are involved, albeit that familial occurrence has been reported only once.[6] Genetic testing, however, is not yet possible. Males are slightly more frequently affected than females in the ratio 1.35 to 1. The incidence of the syndrome is between 1 : 65,000 and 1 : 200,000.[2,6]

The clinical presentation depends on the intracardiac and associated malformations. When a left ventricular diverticulum is isolated, it can manifest itself in different ways. Symptoms such as palpitations, discomfort, and cardiac failure are reported in from two to four-fifths of cases. Abnormal heart sounds are heard in half, chest radiography is abnormal in three-fifths, and electrocardiographic abnormalities are found in the majority, including ventricular tachycardia.[4,7]

Pathologic examination reveals that the diverticulum originates from the left ventricular apex in four-fifths, and from the free left ventricular wall in the remainder. Only rarely do diverticulums originate from the right side.[4,8] Multiple diverticulums are also rare.[9]

The diagnosis of a left ventricular diverticulum can be difficult due to abnormalities of cardiac position, but with echocardiography and angiography, one should be able to establish the diagnosis. Magnetic resonance imaging may be helpful in relating the diverticulum to its adjacent structures. Spontaneous rupture, arrhythmias, and thrombogenicity have all been reported.[4,7] Thus, resection of the diverticulum seems to be indicated.[2,4,8] The timing of resection is still a point of discussion. Some authors prefer intervention as soon as possible,[2] while others advocate delay until the patients become symptomatic.[9]

The prognosis of Cantrell's syndrome depends mainly on the cardiac malformations found. At the time of Cantrell's report, survival was only 37%. Analysis of patients over the last two decades showed an increase in survival to over 60%.[2]

It is recommended that every patient with an omphalocele or other supra-umbilical midline umbilical wall defect should be screened for Cantrell's syndrome, in particular for anterior diaphragmatic hernia and cardiac malformations. A left ventricular diverticulum when found with other malformations is itself suggestive of Cantrell's syndrome. Since intracardiac malformations are then very likely, further analysis is always indicated.

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