

Unusual case of unrepaired omphalocele and uncorrected tetralogy of Fallot in a young adult

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ABSTRACT

An omphalocele is a congenital defect that affects the development of the abdominal wall in the umbilical region, resulting in a hernial-type sac of variable size. The condition is usually diagnosed prenatally and corrected in early infancy to prevent rupture of the covering membranes, which carries a high mortality and morbidity rate. Tetralogy of Fallot is the most common cyanotic congenital heart defect during infancy that is associated with this condition. Most patients experience cyanosis at birth and die in childhood if there is no surgical intervention. Overall, it is uncommon for untreated patients with both omphalocele and tetralogy of Fallot to survive into adulthood. We report the rare case of a 17-year-old young adult with untreated omphalocele and uncorrected tetralogy of Fallot.

Keywords: omphalocele, tetralogy of Fallot, young adult

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INTRODUCTION

An omphalocele represents an embryological defect of the umbilical ring and medial segments of the two lateral abdominal folds. The incidence of omphalocele is reported to be between 1:3,000 and 1:10,000 births.⁽¹⁾ Many anomalies coexist in this condition, including cardiac, gastrointestinal, genitourinary, musculoskeletal and central nervous system defects. Cardiovascular anomalies are most commonly found in patients with epigastric midline omphaloceles, and tetralogy of Fallot is by far the most common. Omphalocele is generally treated with surgery at birth, either as an emergency procedure or a planned procedure, depending on the case and the malformations associated with it. Late presentation of unrepaired omphalocele in adulthood is rare, with only two cases of adult omphalocele reported so far. The first case was a 38-year-old woman with untreated omphalocele and duplication of the inferior vena cava (IVC) and a malpositioned right kidney.⁽¹⁾ The

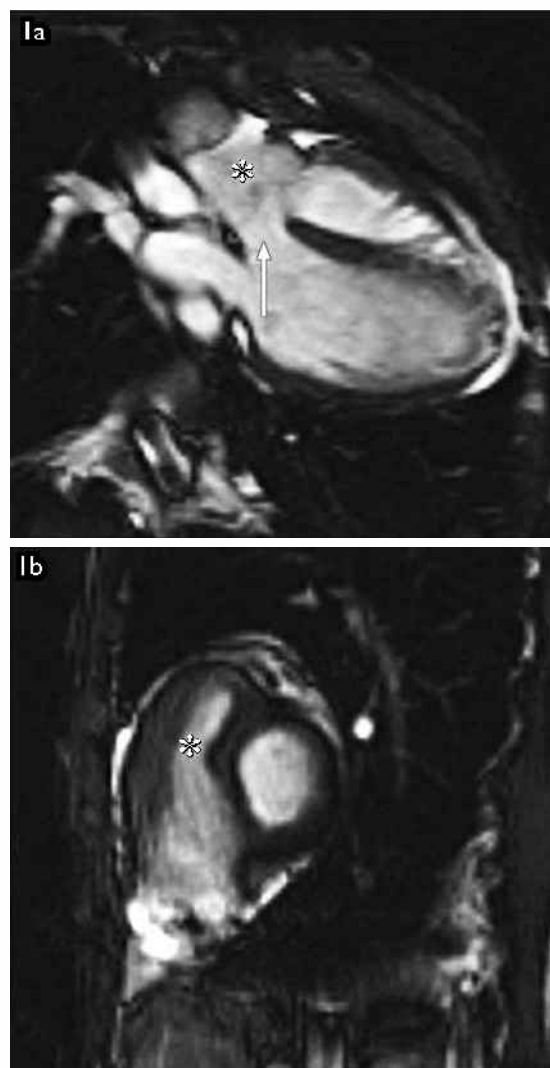


Fig. 1 Steady-state free precession cardiac MR cine still image of (a) the left ventricular outflow tract in end-diastole of the patient with tetralogy of Fallot shows an over-riding aorta (*) and an outlet ventricular septal defect (arrow); and (b) the right ventricular outflow tract in end-systole shows an infundibular hypertrophy (*), but with no significant stenosis.

second was a 29-year-old woman who had a partially treated omphalocele in childhood, with no associated anomalies.⁽²⁾

We describe the case of a 17-year-old young adult who presented with an omphalocele, tetralogy of Fallot, duplication of the IVC and a malpositioned spleen. To the

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best of our knowledge, this is the first case of a young adult with an unrepaired omphalocele and uncorrected tetralogy of Fallot.

CASE REPORT

An asymptomatic 17-year-old male Burmese (New York Heart Association Class I⁽⁶⁾) was referred to our hospital for repair of tetralogy of Fallot. He has a midline abdominal mass from birth, which has been slowly growing over time. On inspection, a bulging lump with deformity of the upper anterior abdomen with thinned-out overlying skin was noted. The lump was non-pulsatile and moved with respiration. On palpation, the mass was firm in consistency and not reducible.

Cardiac magnetic resonance (MR) imaging was performed, which revealed an overriding aorta, ventricular septal defect (VSD), hypertrophied right ventricle with a dysplastic and stenosed pulmonary valve, in keeping with tetralogy of Fallot (Fig. 1). The main pulmonary artery was small in size, with normal calibre of the branch pulmonary arteries. Echocardiography revealed bidirectional flow across a large VSD and an overriding aorta. Abdominal computed tomography (CT) was performed to evaluate the epigastric mass. The localiser showed an opacity projecting over the epigastric-mesogastric region (Fig. 2). Axial CT of the abdomen showed a 9-cm separation between the internal margins of the rectus abdominus muscles and a hernia in the epigastric-mesogastric region (Fig. 3). The liver (segment IV) was herniating through the defect, which was only covered by skin. There was also duplication of the IVC.

The patient was admitted for total correction of tetralogy of Fallot. Intraoperatively, he was found to have severe annular and valvular pulmonary stenosis and a narrowed main pulmonary artery, which corresponded to the cardiac MR imaging findings. Repair of tetralogy of Fallot was done with patch closure of the VSD and right ventricular outflow patchplasty with autologous pericardium. The postoperative period was uneventful. Open repair of the omphalocele with component separation and mesh was performed three years later when the patient was 20 years old. His postoperative recovery at this time was also uneventful.

DISCUSSION

Omphalocele represents an embryological defect of the umbilical ring and medial segments of the two lateral abdominal wall folds that form the anterior abdominal wall during foetal growth.⁽⁴⁾ The aetiology of omphalocele is unknown, but various theories have been



Fig. 2 CT scanogram shows a soft tissue opacity projecting over the epigastric-mesogastric region (arrows).

postulated. These include failure of the bowel to return to the abdomen by 10–12 weeks, given that physiological umbilical herniation should not persist beyond 12 weeks. After 12 weeks of gestation, one must consider omphalocele as the cause of a mass in the base of the umbilical cord and beyond the confines of the abdominal wall.⁽⁵⁾

The size of the defect is variable and can range from 4 cm to more than 10 cm.⁽⁴⁾ Omphaloceles can be divided into two groups depending on the size of the hernial defect; minor and major (giant). A minor omphalocele occurs when the defect is 4 cm or less. A major or giant omphalocele is classified as a 5 cm or larger defect.⁽⁶⁾ The hernial sac may contain small and large bowel, stomach, liver, spleen, urinary bladder, uterus and ovaries. Our patient had a giant omphalocele containing the left hepatic lobe, which was seen on multiplanar CT of the abdomen.

Omphaloceles can also be subdivided according to the site of the defect into epigastric (classic omphalocele) with cephalic fold defect, central type with a lateral wall defect greater than 4 cm and the hypogastric/caudal type with caudal fold defect.⁽⁴⁾ Chromosomal anomalies are common (accounting for 40%–60%), and these include trisomies 13, 18 and 21 as well as Turner, Klinefelter and triploidy syndromes.^(4,7) Associated cardiovascular anomalies are seen in up to 50% of patients with omphaloceles. However, the frequency of congenital

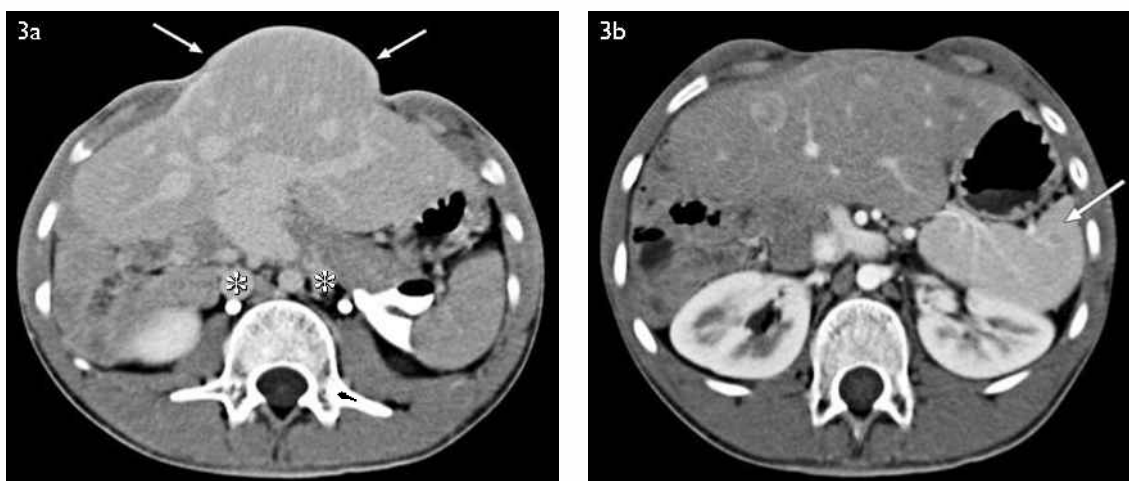


Fig. 3 Contrast-enhanced CT images show (a) diastasis of the rectus abdominus muscles with a partially herniated malformed liver covered by a thin layer of skin (white arrows) and double infrarenal inferior vena cavae (*); and (b) a malpositioned spleen (arrow).

heart disease reaches up to 80% in patients with a middle caelosomia, which includes the liver, as in the case of our patient.⁽⁸⁾ Tetralogy of Fallot is by far the most common associated cardiac anomaly (33%).⁽²⁾ An association between anomalous vena cavae and omphalocele has been described earlier, with suggestion of a common aetiology.⁽⁸⁾ Other anomalies include genitourinary (40%), respiratory, craniofacial, gastrointestinal and diaphragmatic anomalies.

Diagnosis of omphalocele can be easily made by prenatal ultrasonography. A definitive diagnosis of omphalocele is possible only beyond 12 weeks of gestation, when confusion with physiologic midgut herniation is no longer possible. A number of sonographic features differentiate an omphalocele from physiologic midgut herniation. For example, a midgut herniation seldom exceeds 7 mm in diameter, whereas omphaloceles are much larger, sometimes even larger than the abdomen. In addition, midgut herniation seldom persists after 12 weeks of gestation or in a foetus with a crown-rump length measurement of more than 44 mm. MR imaging studies of the foetus may help in corroborating and refining the ultrasonographic diagnosis of complex foetal defects.⁽⁵⁾

Methods of managing omphalocele patients include non-operative treatment in cases of large or giant omphaloceles, which involves application of silver sulfadiazine (topical antimicrobial drug) or Aquacel gel (ConvaTec, Skillman, NJ, USA) to toughen the sac and protect it from infection. Surgery is postponed in these cases in order to allow the abdominal cavity to enlarge naturally as the baby grows.^(9,10) Primary operative repair methods include direct fascial closure for defects of up to 6 cm and the use of alloplastic material fixed to the

fascia to fill the gap. The definitive treatment for tetralogy of Fallot is surgery. Previously, palliative procedures that allowed for increased pulmonary flow and thus, decreased cyanosis were performed e.g. Blalock-Taussig shunt. Currently, it is recommended that the patient undergo complete surgical repair with closure of the VSD and relief of the right ventricular outflow tract obstruction at a very early age.^(11,12) Without surgical repair, most patients with isolated tetralogy of Fallot would die in childhood. In the past, survival without surgery was 66% at one year of age, 40% at three years of age, 11% at 20 years of age and 3% at 30 years of age.⁽¹³⁾

Prognosis depends on the presence and severity of associated anomalies. Isolated omphalocele is believed to have relatively good prognosis with a higher rate of survival than if accompanied by a major structural or karyotype abnormality. It is unusual for a patient to survive into young adulthood without repair of either the omphalocele or tetralogy of Fallot, as the prognosis for infants with both omphalocele and congenital heart disease is generally poor, with an 80% mortality rate compared to a 30% mortality rate for infants with omphalocele but without congenital heart disease.⁽²⁾ In patients with combined omphalocele and cardiac anomalies, multidisciplinary consultation between the cardiologist, radiologist, cardiac surgeon, general surgeon, intensivist and anaesthetist would best determine the treatment options in consultation with the patient and his family.

In conclusion, omphalocele is known to be associated with tetralogy of Fallot. It is, however, unusual to see a young adult patient with untreated omphalocele and uncorrected tetralogy of Fallot, as both conditions carry high mortality rates. To the best of our knowledge,

this is the first case in the literature in which a young adult patient has both an untreated omphalocele and unrepaired tetralogy of Fallot.

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