Pentalogy of Cantrell

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الخلاصة :

متلازمة (خماسية) كانترل من الحالات المرضية النادرة والتي تظهر بشكل متلازمة تشوهات خلقية ولادية تصيب واحد من كل مليون ولادة حية وتشتمل على تشوه جدار البطن وبروز الاحشاء ,تشوه العظم القصي ,فتق الحجاب الحاجز , بروز عضلة القلب مع شغاف القلب وتشوه القلب. دراسة لحالة طفل ,ذكر حي بلغ من العمر يومان ادخل الى وحدة جراحة الاطفال في مستشفى الولادة والاطفال التعليمي في الديوانية وكان مصاب بما اشتمل عليه تعريف المتلازمة المذكورة , تشوه جدار البطن الخارجي مع بروز احشاء البطن(الكبد ,الطحال وجزء كبير من الامعاء الغليظة) ,تشوه العظم القصي , بروزشغاف القلب وفتق الحجاب الحاجز. اجريت له كافة اساسيات انعاش الطفل وكان القرار للتداخل الجراحي على مرحلتين لتلافي المضاعفات التي قد تحصل من أي تداخلات جراحية تنشأ من جراحة واحدة لتصليح هذا التشوه واجريت له الجراحة الاولى بعد يومين على دخوله الى ردهة الجراحة وتم تصليح تشوه جدار البطن والصدر وفتق الحجاب الحاجز واجريت له الجراحة الثانية بعد سنتين من الجراحة الاولى لوضع شبكة (داكرون) لتقوية جدار الصدر والبطن ومنع فتق احشاء الصدر او البطن الداخلية.

ABSTRUCT Pentalogy of Cantrell is a rare syndrome that consists of defects involving the diaphragm, abdominal wall, pericardium, heart and lower sternum. A 2 –day- old male neonate presented to the pediatric surgical unit in the Maternity and Child Teaching hospital\AL-diwaniya with abdominal wall defect,precious baby deliverd by C\S to the mother with history of primary infertility for more than 13 year, The first clinical evaluation revealed epigastric omphalocele (liver ,spleen and transverse colon were exposed with rupture sac , sternal defect , and ectopia cordis. U\S ,echocardogram and radiological studies were done to exclude others associated anomalies , Staged closure decided because the primary closure will significantly compromise ventilation or abdominal viscera .The first surgery was closure of both abdominal and thoracic defect ,bilateral flank and lateral chest wall release incisions and skin flap done for this closure. The patient had an uncomplicated post-operative course. A chest CT was performed at 2 weeks of life and demonstrated a very hypoplastic appearing anterior chest wall and a stenosis of the left mainstem bronchus as the cause of the patient's left lung hyperinflation. Monthly evaluation were done till he became at age

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of two years and every thinks were normal a part from abdominal distention (herniated viscera) which affected and limited the physical activity so the second stage was done, reinforced Dacron mesh placed to the anterior abdominal wall to protects the herniated bowel and redirects the pressure in the abdomen and to promotes enlargement of abdominal cavity.

INTRODUCTION

Pentalogy of Cantrell is a rare syndrome that consists of defects involving the diaphragm, abdominal wall, pericardium, heart and lower sternum [7].

The syndrome consists of:

- Omphalocele
- Anterior diaphragmatic hernia
- Sternal cleft
- Ectopia cordis
- Intracardiac defect: ventricular septal defect or diverticulum of left ventricle

The differentiation of Cantrell's pentalogy form other abdominal wall defects is extremely important.The differential diagnosis includes isolated thoracic ectopia cordis,amniotic band syndrome and body stalk anomalies.The key features for distinguishing these conditions is the position of abdominal wall defect in relation to the umbilical cord insertion,eviscerated organs,the presence or absence of membranes or bands,and associated anomalies.Omphalocele in Cantrell's pentalogy usually involves a midline defect at the umbilical cord insertion.An eccentric large lateral defect and adherence of the placenta to the defect are typically present in body stalk anomalies.The presence of an unexplained ventral wall defect along with extremity deformity with an adherent band suggests amniotic band syndrome.

Carmi et al. (1990) suggested X-linked dominant inheritance for a previously undescribed malformation syndrome. The features were diaphragmatic and ventral hernias, hypoplastic lung, and cardiac anomalies such as transposition of the great vessels and patent ductus arteriosus.

Carmi et al. (1990) pointed out that another syndrome that combines defects of the abdominal wall and the diaphragm is the sporadic pentalogy of Cantrell (Cantrell et al., 1958) [1,2,3]. Although the abdominal wall defect, even in its severe form, was equally present in females and males, the diaphragmatic and lung anomalies were mostly confined to males. In 7 of 10 affected males, these anomalies were fatal. Cardiovascular abnormalities were not diagnosed in any living affected member of the family. Carmi et al. (1990) described a family in which 4 male fetuses had cystic hygroma, cleft palate, omphalocele, or diaphragmatic hernia in various combinations [4,5]. Toriello and Higgins (1985) described a kindred in which 5 males in as many sibships, related through women, had 6 different defects which were all either midline or midline-associated malformations: hydrocephalus, anencephaly, cleft lip, congenital heart defect, renal agenesis, and hypospadias. Toriello and Higgins (1985) postulated that the midline may be a developmental field as suggested by Opitz and Gilbert (1982), and that a single gene mutation, in this instance on the X chromosome, had disrupted development [7.10]. Carmi and Boughman (1992) ascertained 5 cases of pentalogy of Cantrell through the Baltimore-Washington population-based study of infants with congenital cardiovascular malformations--a regional prevalence of 5.5 per 1 million liveborn infants. Three of the patients had cleft lip with or without cleft palate. Carmi and Boughman (1992) again proposed this as evidence of a ventral midline developmental field. At least 2 of the 5 patients were female [3,4]. Martin et al. (1992) indicated that the association of sternal fusion defects with various cardiac, diaphragmatic, and anterior body wall defects represents a developmental field complex that includes the pentalogy of Cantrell and ectopia cordis. They presented a family in which 3 consecutively born brothers had extensive diaphragmatic defects; 2 of the brothers had the pentalogy of Cantrell and 1 of these 2 also had ectopia cordis [10].

In studies of an extended family including 14 affected individuals, Carmi et al. (1993) obtained positive lod scores with markers in the region Xq22-q27. The maximum lod score was obtained for linkage with HPRT at Xq26.1; maximum lod = 5.11 at theta = 0.042. Additional results

indicated that the TAS gene is located between the DXS425 and HPRT loci (Xq25-q26.1). Parvari et al. (1994) obtained a multilocus lod score of 12.4 when the linkage analysis utilized additional markers in Xq25-q26. Using microsatellite polymorphic markers, Parvari et al. (1996) narrowed the region of the TAS gene to an interval of about 2.5 Mb [8,9].

Management includes a complete workup including karyotype and complete ultrasonographic search for other anomalies performed.Termination of pregnancy can be offered before viability.After viability ,periodic ultrasonographic evaluations of the lesions,fetal growth and delivery in a tertiary centre is recommended.Cesarean section delivery does not nessecarily improve the prognosis [3,7].

Case report

A 2 –day- old male neonate presented to the pediatric surgical unit in the Maternity and Child Teaching hospital\AL-diwaniya with abdominal wall defect, precious baby delivered by C\S to the mother with history of primary infertility for more than 13 year(picture 1) .

All the principles of immediate postnatal management in the neonate were started in pediatric surgical unit and directed at correcting hypovolemia , preventing hypothermia and monitoring for sign of sepsis. The viscera were wrapped in sterile guze , the neonate placed in warmer ,N\G tube placed to decompress the bowel. Fluid resuscitation started , bladder catheter was placed to monitor urine output and intravenous fluid administration was adjusted to achieve adequate urine output ,tissue perfusion, pulse and blood pressure.

The first clinical evaluation revealed epigastric omphalocele (liver ,spleen and transverse colon were exposed with rupture sac , sternal defect , and ectopia cordis. U\S ,echocardogram and radiological studies were done to exclude others associated anomalies. A chest and abdominal film from the third day of life demonstrated the ectopia cordis and a paucity of bowel gas in the abdomen. Abdominal US from the third day of life demonstrated massive ascites, and no hepatic lesions were seen. An echocardiogram noted the presence of double outlet right ventricle.

On the fourth day of life the patient was taken to the operating room for closure of the midline defect. The midline defect extended from just below the umbilicus to halfway up the heart and was 10-12 centimeters wide. The omphalocele sac was excised revealing the apex of the heart

and the liver. The pericardium was intact.. The omphalocele was closed by approximating the abdominal skin. The heart was covered with skin.

Staged closure decided because the primary closure will significantly compromise ventilation or abdominal viscera .

The first surgery was closure of both abdominal and thoracic defect ,bilateral flank and lateral chest wall release incisions and skin flap done for this closure (picture 2).

The patient had an uncomplicated post-operative course. A chest CT was performed at 2 weeks of life and demonstrated a very hypoplastic appearing anterior chest wall and a stenosis of the left mainstem bronchus as the cause of the patient's left lung hyperinflation.
Monthly evaluation were done till he became at the age of two years and every thinks were normal a part from abdominal distention (herniated viscera) which affected and limited the physical activity so the second stage was done, reinforced Dacron mesh placed to the anterior abdominal wall to protects the herniated bowel and redirects the pressure in the abdomen and to promotes enlargement of abdominal cavity (picture 3).
After three months from the second surgery the family visited me ,the child was in normal life a part from attack of constipation and anal fissure!.

Discussion

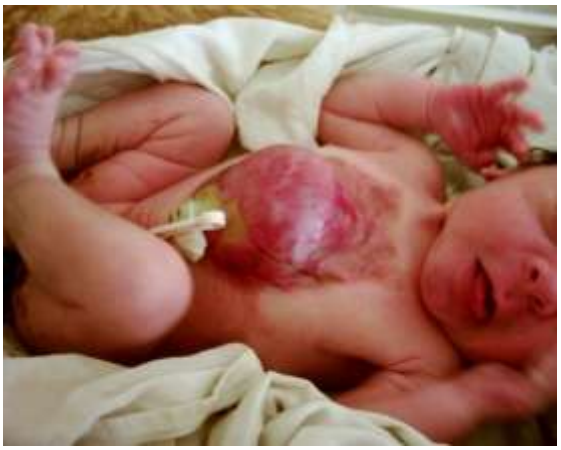
The differentiation of Cantrell's pentalogy form other abdominal wall defects is extremely important .The differential diagnosis includes isolated thoracic ectopia cordis,amniotic band syndrome and body stalk anomalies .The key features for distinguishing these conditions is the position of abdominal wall defect in relation to the umbilical cord insertion, eviscerated organs, the presence or absence of membranes or bands,and associated anomalies . Omphalocele in Cantrell's pentalogy usually involves a midline defect at the umbilical cord insertion .An eccentric large lateral defect and adherence of the placenta to the defect are typically present in body stalk anomalies.The presence of an

unexplained ventral wall defect along with extremity deformity with an adherent band suggests amniotic band syndrome.

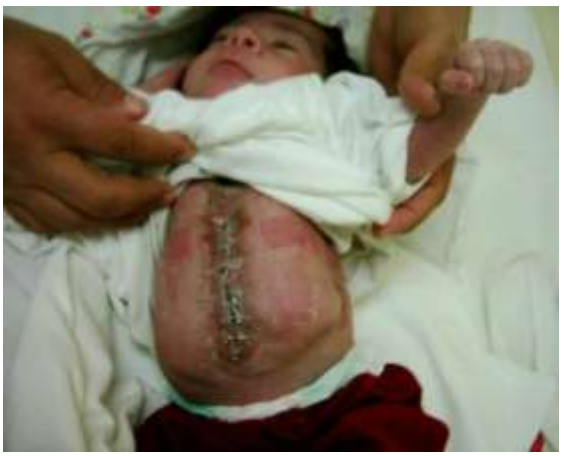
Management includes a complete workup including karyotype and complete ultrasonographic search for other anomalies performed.Termination of pregnancy can be offered before viability. After viability ,periodic ultrasonographic evaluations of the lesions, fetal growth and delivery in a tertiary centre is recommended. Cesarean section delivery does not nessecarily improve the prognosis.

Mild cases of Cantrell's pentalogy can be repaired in a single stage. In our case we, advocate a two-stage repair. The goals of the first operation are to provide soft tissue coverage to the abdomen and heart by making space in the posterior mediastinum. The pericardioperitoneal communication is closed either primarily or by graft. In some occasions it is not possible to reduce the abdominal contents and the heart because hemodynamic instability. In these cases the heart can be reduced posteriorly, the diaphragmatic defect approximated after the intestines and liver are removed from the chest, and a the skin of the abdominal wall approximated for progressive reduction. Hemodynamics tolerate slow reduction the best. And here we facing a real problem how to allow the chest cavity to grow and make space for the heart!. The heart is returned to the left or right pleural spaces depending on where the apex points, and wheter mesocardia, levocardia, or dextrocardia is exhibited. The chest wall is reconstructed if necessary by means of a neosternum formed by the ribsand perichondrium which is still unavailable in our hospital so we decided to cover the heart by skin only.

Location and route of delivery can then be chosen to facilitate postnatal management. Initial treatment consists of coverage of the viscera and fluid resuscitation. Several options are then available for definite surgical management, depending on the size of the defect, the condition of the underlying viscera, and the status of the baby. Meticulous and systematized therapeutic management is needed for the favorable outcome of the patient .The principles of surgical reconstruction are based on the anatomy of the defect, associated structural abnormality, and the age of the patient .The successful surgical outcome in this baby is the result of continually evolving surgical and neonatal care in our part of the world.



Picture 1



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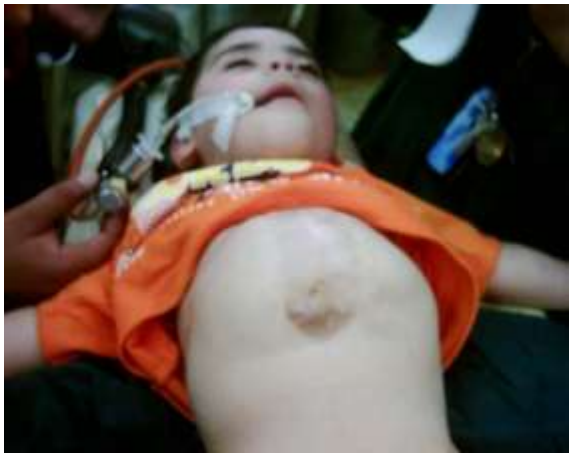

Picture 3

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