J Ped Surg Case Reports 3 (2015) 476–480

Contents lists available at ScienceDirect

Journal of Pediatric Surgery CASE REPORTS

journal homepage: www.jpscasereports.com

Pentalogy of Cantrell with thoracoabdominal ectopia cordis: Attempted surgical correction and review of recent literature to aid prognostication prior to surgery

Vered Lack [a], Christina Oetzmann von Sochaczewski [a,*], Krubin Naidoo [b], Jerome Loveland [a]

[a] Department of Paediatric Surgery, Chris Hani Baragwanath Academic Hospital, Johannesburg, South Africa
[b] Department of Cardiothoracic Surgery, Charlotte Maxeke Johannesburg Academic Hospital, University of the Witwatersrand, Johannesburg, South Africa

ARTICLE INFO

Article history:
Received 26 August 2015
Accepted 1 September 2015

Key words:
Pentalogy of Cantrell
Thoracoabdominal ectopia cordis
Thoracic ectopia cordis
Extrathoracic heart
Congenital heart disease

ABSTRACT

Complete ectopia cordis (EC) is a rare congenital condition where the heart is extrathoracic, uncovered by pericardium and skin. This condition is often accompanied by intracardiac as well as other congenital anomalies. The thoracoabdominal type is frequently associated with varying degrees of Pentalogy of Cantrell (POC). Unless addressed early post-natally, EC is incompatible with life. Surgery is usually staged, with cover of exposed organs an immediate priority, followed by repair of structural cardiac lesions. Formal repair of the chest wall is performed at a later stage. We present a case of thoracoabdominal EC with complete POC. The patient underwent surgery to cover the exposed heart and the abdominal wall defect, but demised within 48 h due to systemic sepsis. Data gained from a literature survey shows that if the congenital heart defect is haemodynamically stable, and if cover is achieved immediately after birth, there is an associated improved survival. In addition, antenatal diagnosis was found to improve planning of delivery, resulting in improved hemodynamic stability, decreased incidence of sepsis, shorter time to intervention and better overall survival.

EC is defined as a heart which is congenitally displaced outside the thoracic cavity, either partially or completely [1,2]. It was noted as early as 5000 years ago by the Babylonians [2,3], as documented in the cuneiform tablets of Nineveh [4]. Today, the incidence is 5.5–7.9 per million live births and without surgical intervention is incompatible with life [5]. To date, surgery has met with modest success. We present a case of Class 1 POC with complete thoracoabdominal EC, discuss the first stage of surgical management and review the literature with respect to antenatal diagnosis, mode of delivery, time to operation, degree of intracardiac anomalies and available options for surgical cover. Thereafter the results are analyzed.

1. Background

EC was first mentioned in 1671 by Niels Stensen in his description of the first case of Tetralogy of Fallot (TOF), which also happened to be a case of EC [6]. It was later independently described by Haller and Martinez [7]. Between 1818 and 1962 "ectopia cordis" was the subject of various classifications. Weese classified EC into three anatomical groups: *ectopia cordis suprathoracica, ectopia cordis cum sterni fissure* and *ectopia cordis subthoracica*. Townsend used the terms "cervical heart," "pectoral heart with fissure of the sternum" and "abdominal heart with a defect in the diaphragm." In 1948 Byron added a fourth group, the "thoracoabdominal heart," but in 1962 Kanagasuntheram and Verzin pointed out that these subdivisions do not cover all the variations of the defect and suggested the following classification: 1) Cervical: the heart is entirely in the cervical region but the sternum is intact. 2) Thoracocervical: the heart is partially in the cervical region but the cranial end of the sternum is defective. 3) Thoracic: the sternum is defective and the heart lies partially or completely outside the thorax. 4) Abdominal: the diaphragm is defective, allowing the heart to enter the abdominal cavity. 5) Thoracoabdominal: several defects are present [7]. The heart usually lies in normal relationship to the other thoracic structures, although there may be some degree of dextrorotation [8].

* Corresponding author. Tel.: +27 84 6732356.
E-mail address: c.oetzmann@gmail.com (C. Oetzmann von Sochaczewski).

http://dx.doi.org/10.1016/j.epsc.2015.09.001

EC is further described as complete or partial. In partial EC the heart can be seen to pulsate through the skin or may have complete pericardial cover, whereas in complete EC the naked heart is displaced outside the thoracic cavity and is without pericardium [3].

The thoracoabdominal type was invariably found to be associated with defects in the supra-umbilical abdominal wall, lower sternum, anterior diaphragm, and pericardium, as well as the associated intracardiac defects [7].

This consistent cluster of lesions was described by Cantrell as a predictable pentalogy in 1958, and is referred to by some as Cantrell-Haller-Ravitch Syndrome [9]. The incidence of isolated POC is reported to be 5.5 per million live births, with the complete spectrum of lesions presenting infrequently [10]. For this reason, in 1972, Toyama suggested a classification for POC as follows: Class 1: definite diagnosis, with all five defects present; Class 2: probable diagnosis, with four defects present, including intracardiac and ventral wall abnormalities, and Class 3: incomplete expression, with various combinations of defects present, including a sternal abnormality [11].

Co-existent abnormalities may adversely affect prognosis. These include congenital heart defects such as ventricular septal defect, atrial septal defect (ASD), TOF, left ventricular diverticulum as well as other more complex lesions, including double outlet right ventricle (DORV) or transposition of the great vessels (TGV), pulmonary hypoplasia, craniofacial malformations, chromosomal abnormalities, spina bifida, limb abnormalities, scoliosis and others [10,12].

POC is reported to have a male propensity, with females demonstrating more severe associated malformations and poorer prognosis [12]. Occasional cases have been associated with chromosomal abnormalities such as triploidy, and a weak familial X-linked inheritance has been suggested [12].

2. Case report

A 2080 g female born at 35 weeks gestation by normal vaginal delivery was transferred to our facility on day two of life with a provisional diagnosis of EC (Video A Supplemental Data). She was born to a 20-year-old primigravida without prior obstetric history who was positive for Rhesus antigen and Human Immunodeficiency Virus (HIV) negative. There was no consanguinity between the parents and no family history of congenital cardiac malformations. Delivery was an uncomplicated vertex presentation with APGAR scores of 7 and 9 at 1 and 5 min respectively. The baby was placed on nasal prong oxygen at 2 L/minute immediately after birth and the defect covered with transparent plastic sheeting. Transfer to the tertiary hospital facility took 5 h. On arrival the patient was haemodynamically stable. There was a large 6 × 4 cm thoracic and supra-umbilical abdominal wall defect with heart and great vessels exposed. These lacked skin or pericardial cover (Fig. 1). In addition there was a small omphalocele and an anterior diaphragmatic defect. The sternum was absent. Auscultation revealed equal air entry bilaterally. The anus was patent and meconium had been passed. Documented dysmorphic features included hypertelorism, microphthalmia, micrognathia, and a flattened nasal bridge. Limbs appeared normal. A chest X-ray was done (Fig. 2), and echocardiography demonstrated an ASD measuring 5 mm. Features were consistent with a diagnosis of Class 1 POC, as per Toyama's definition, with a complete thoracoabdominal EC.

Although the infant was phenotypically dysmorphic, results of genetic testing were not available, and on account of the child's stable condition, repair of the defect was planned.

The size of the defect was thought to preclude cover with native tissue without causing compression of the heart, thus artificial materials were selected for initial cover and to offer a scaffold for pericardial regrowth with a view to "pericardial containment." Written informed consent was obtained from the parents.

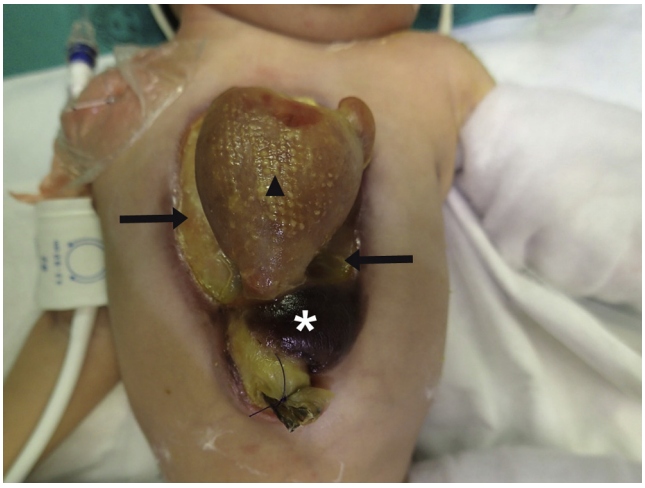

Fig. 1. Complete Pentalogy of Cantrell: Ectopia cordis (triangle), small atrial septal defect, deficient sternum and absent of diaphragmatic pericardium (residual pericardium indicated by arrows), supra-umbilical abdominal wall defect (star) with protruding liver and absent of anterior diaphragm.

2.1. Surgical method

On day four of life, surgery was undertaken by a team comprising pediatric surgeons and pediatric cardiac surgeons. The sac covering the omphalocele was opened and excised, defining the abdominal defect, after which this mobilization was extended separating the heart from the thoracic wall. The sheath and skin were separated, and the umbilical stump was trimmed to create a smooth contour for the abdominal defect. The first patch (CorMatrix ECM Technology 7 × 10 cm) was fashioned to fit the contour of the heart taking care not to create tension. This was sutured to the rudimentary posterior pericardium using continuous 7-0

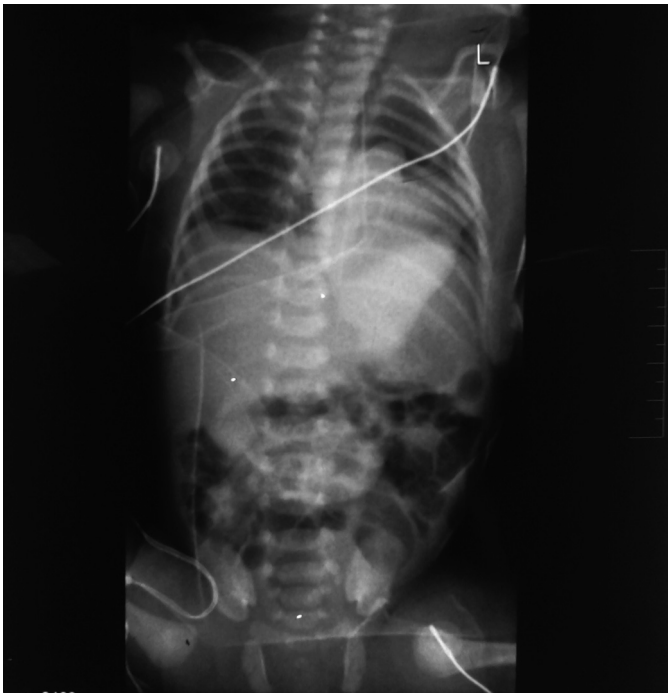

Fig. 2. Plain thoracoabdominal film showing abnormal heart position, signs of right ventricular hypertrophy, bilaterally reduced pulmonary vasculature, central abdominal mass containing bowel compatible with omphalocele. Rotation to the left.