First reported case of pentalogy of Cantrell in Sri Lanka

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Introduction

Pentalogy of Cantrell (POC) is a rare syndrome first described by Cantrell, characterised by the presence of five major congenital defects involving the diaphragm, abdominal wall, diaphragmatic pericardium, lower sternum and multiple congenital intracardiac abnormalities[1]. POC has a male dominance with the commonest outcome being early neonatal death[2]. Although early and aggressive surgical treatment is recommended, prognosis depends on the size of the abdominal wall defect, the type of the ectopia cordis and associated anomalies[3]. Ectopia cordis (EC) was first described by Haller in 1706 as complete or partial displacement of the heart outside the thoracic cavity[2]. It is a rare congenital defect in fusion of the anterior chest wall[3]. In the most common form of ectopia cordis, the sternum is split, and the heart protrudes outside the chest[2]. In other forms, the heart protrudes through the diaphragm into the abdominal cavity or may be situated in the neck[2]. Ectopia cordis is commonly associated with intracardiac anomalies[4].

Case report

A baby boy was born to an 18-year-old primigravida mother and 25-year-old father and the mother was transferred from a district hospital to De Soysa Hospital for Women, Colombo after antenatal detection of EC in the absence of other antenatal complications. Baby was delivered via emergency lower segment caesarean section at 34 weeks of gestation due to cardiotocography changes with a birthweight of 1.850 kg (between median to -1SD), birth length of 43cm (between median to -1SD) and head circumference 32cm (median to +1SD). Apgar score was 10 at 1, 5 and 10 minutes of age.

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Examination revealed an externally placed heart covered with skin over the anterior chest wall (Figure 1).

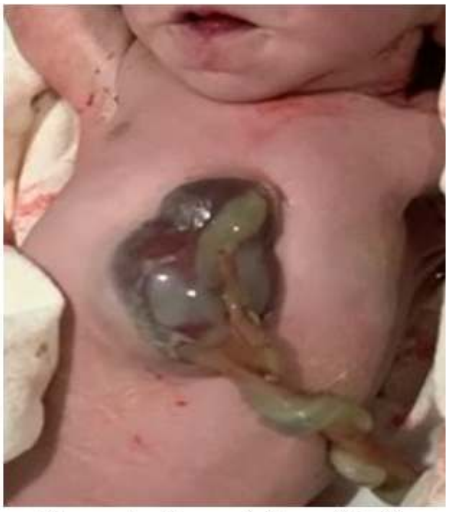

Figure 1: ***Presentation at birth***

Baby was haemodynamically stable with a heart rate of 168 beats per minute, capillary refill time of less than 2 seconds and a mean arterial pressure of 40mmHg. Rest of the examination did not reveal any abnormality.

Tables 1 and 2 show the investigations carried out in the patient.

No abnormality was detected on full blood count, C-reactive protein, serum electrolytes, blood culture, ultrasound scan of the brain, ultrasound scan of the abdomen and kidney, ureter and bladder (KUB) x-ray study.

While Figure 2 shows the 3 D constructed anterior computed tomography (CT), Figure 3 demonstrates the coronal view.

2D echocardiogram done on day one showed a large muscular ventricular septal defect (VSD), large ostium secundum atrial septal defect (OS-ASD), tiny patent ductus arteriosus (PDA), slightly small aortic arch and hyperkinetic pulmonary arterial hypertension. 2D echocardiogram done on day 7 confirmed the same findings. A pus collection over the skin covering the heart, noted on day 3, resolved after treatment with intravenous flucloxacillin for 5

days followed by oral cloxacillin for another 5 days (due to unavailability of oral flucloxacillin in the hospital setting at the time). Contrast enhanced CT scan of the chest and abdomen, done on day 3 of life, revealed a midline defect in the epigastrium just below the sternum. It also demonstrated herniation of left ventricle with small thoracic cavity dimensions along with anticlockwise rotation of the heart. In addition, there was coarctation of the aorta before the origin of the left subclavian artery without any pre or post stenotic dilatation in addition to intracardiac shunts in the form of VSD and ASD. Furthermore, it demonstrated herniation of few bowel loops along with segment IV and II of the liver. These findings confirmed Pentalogy of Cantrell with ectopia cordis in conjunction with defects in the sternum, anterior diaphragm and supraumbilical abdominal wall resulting in herniation of the left ventricle, liver, and bowel loops with evidence of intracardiac shunts and coarctation of aorta.

Table 1: ***Investigations carried out in the patient***

Investigation	Day 1	Day 14
Total white blood cell count (x 10^9/L)	10.82	10.56
Neutrophil %	53.2	37.7
Haemoglobin (g/dL)	16.6	13.5
Platelet count (x 10^9/L)	296	547
C-reactive protein (mg/L)	1.6	1.01
Blood urea (mg/dL)		9.4
Serum creatinine (µmol/L)		31.7
Total bilirubin (mg/dL)	11.0	6.83
Serum sodium (mmol/L)	145.0	136.5
Serum potassium (mmol/L)	5.78	4.89
Prothrombin time (seconds)	15.2	
International normalised ratio (INR)	1.486	
Activated thromboplastin time (seconds)	26.8	

Table 2: ***Special investigations carried out in patient***

Special Investigation	Day of life	Findings
2D echocardiogram	Day 1 and Day 7	Ectopia cordis having large muscular ventricular septal defect, large ostium secundum atrial septal defect, tiny patent ductus arteriosus, slightly small left aortic arch and hyperkinetic pulmonary artery hypertension; systemic to pulmonary communication with increased pulmonary flow via large ventricular septal defect.
Ultrasound scan of brain	Day 10	Normal study
Ultrasound scan of abdomen and kidney	Day 10	No structural anomalies other than herniation of few bowel loops as demonstrated on computed tomography
Blood culture	Day 1	No growth
Skin swab culture	Day 1 and Day 3	No growth

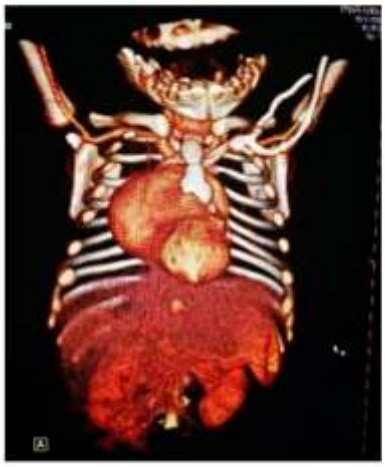

Figure 2: *3D reconstruction of the computed tomography*

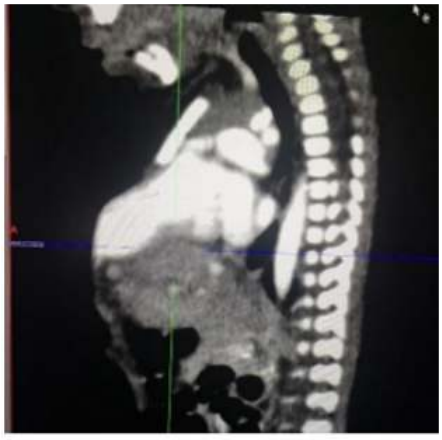

Figure 3: *Coronal view of computed tomography*

Following a multidisciplinary meeting involving paediatric cardiologists and paediatric cardiothoracic surgeons, surgery was considered as a high-risk procedure due to complex nature of VSD and need of arch repair and reconstruction of sternum at the same time. The baby was managed conservatively after counselling both parents, as surgical management was deemed to be extremely high risk with little chances of success. Baby was discharged on day 17 of life with multivitamin, folic acid, and iron after establishing breastfeeding (Figure 4).

Figure 4: ***Baby at 17 days of life***

Baby needed hospital admission due to a lower respiratory tract infection (LRTI) at 3 months of age (Figure 5). Although he recovered from the first LRTI, he succumbed to another LRTI 2 weeks later.

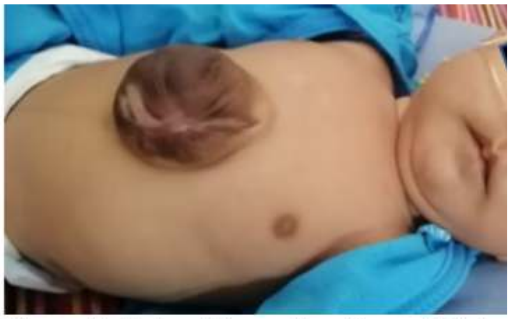

Figure 5: ***Baby at 3 months of age while in hospital for lower respiratory infection***

Discussion

POC results from inappropriate differentiation of a segment of lateral mesoderm between 14-18 days of embryonic life and has an incidence of 5-8 per million live births[1]. It is classified into 3 classes according to the number of components that are present: Occurrence of all 5 defects is known as Class I, occurrence of 4 defects with intracardiac and ventral abdominal wall abnormalities as Class II and incomplete expression of the disorder with sternal anomalies as Class III[1]. Intracardiac anomalies associated with EC, in decreasing prevalence are VSD, ASD, tetralogy of Fallot, a left ventricular diverticulum and pulmonary hypoplasia[1]. This baby demonstrated Class I POC with VSD, ASD, PDA and coarctation of the aorta. Treatment strategies and prognosis depends on the size of the abdominal wall defect, the type of EC and associated anomalies[3]. Although it is recommended that aggressive surgical procedures should be carried out without delay, the overall prognosis is poor[3]. The surgical management of EC primarily involves soft tissue coverage of the heart, replacement of the heart in thoracic cavity, repair of the intracardiac defects and reconstruction of the chest wall[5]. The surgical correction is often difficult due to hypoplasia of thoracic and abdominal cavities and inability to enclose the ectopic heart as in this baby[5]. Most babies with EC die within the neonatal period; especially within the first week of life[5]. Only a few babies thrived well following the corrective surgery whereas most died in the initial post operative period[5]. Our patient survived for 3.5 months until succumbing to a LRTI. This raises the question whether transfer of babies with major congenital anomalies with poor prognosis, justify the cost incurred on the health care system as well as the inconvenience and cost caused to the family.

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References

1. Naburi H, Assenga E, Patel S, Massawe E, Manji K. Class II pentalogy of Cantrell. *BMC Research Notes* 2015; **8**: 318. https://doi.org/10.1186/s13104-015-1293-7
PMid: 26219662 PMCid: PMC4517504

2. Pius S, Ibrahim HA, Bello M, Tahir MB. Complete ectopia cordis: A case report and literature review. *Case Reports in Pediatrics* 2017; **2017**: 1858621. https://doi.org/10.1155/2017/1858621
PMid: 28503337 PMCid: PMC5414485

3. Shad J, Budhwani K, Biswas R. Thoracic ectopia cordis. *BMJ Case Reports* 2012; 2012: bcr1120115241. https://doi.org/10.1136/bcr.11.2011.5241 PMid: 23035158 PMCid: PMC4543290

4. Dias GFIM, Santos AV dos R, Martins CFCP, Ferreira APD, Fonseca JMD da. Ectopia cordis: a case report. *Rev Bras Saude Mater Infant* 2014; **14**(3): 287–90. https://doi.org/10.1590/S15193829201400 0300010

5. Alphonso N, Venugopal PS, Deshpande R, Anderson D. Complete thoracic ectopia cordis. *European Journal of Cardio-Thoracic Surgery* 2003: **23**(3): 426–8. https://doi.org/10.1016/s10107940(02)008 11-4 PMid: 12614821