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Case report/Kazuistyka

The incomplete pentalogy of Cantrell – A case report

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ABSTRACT

We report a case of a female neonate with an incomplete (Class II) pentalogy of Cantrell (PC) presenting: omphalocoele, thoracoabdominal type of partial ectopia cordis with ventricular septal defect and valvular pulmonary stenosis. The patient underwent a successful complete operation. We discuss associated anomalies that might occur with PC and the general overall prognosis for patients with PC. This report describes a very rare case of a patient with PC and coexisting partial ectopia cordis who survived.

Introduction

Pentalogy of Cantrell (PC) is an extremely rare multiple congenital malformation syndrome and was first described in 1958 by James R. Cantrell et al. [1]. It comprises of the following five characteristics: (1) midline, upper abdominal wall disorder (*e.g.* omphalocele, gastroschisis), (2) defect of the lower sternum (*i.e.* cleft sternum or absent sternum), (3) anterior diaphragmatic defect (*i.e.* hypoplastic diaphragm, anterior diaphragmatic hernia), (4) pericardial abnormality (*e.g.* ectopia cordis) and (5) congenital abnormalities of the heart (*e.g.* tetralogy of Fallot, ventricular septal defect, atrial septal defect). The full pentalogy is very rare with a frequency of approximately 1 incidence per 65 000–100 000 live births and because of its different variants, there are also less severe cases described in the literature.

Pentalogy of Cantrell has a very high mortality rate despite significant improvements in neonatal surgery. The prognosis for patients depends mostly on the severity of cardiac malformation with up to 95% mortality when ectopia cordis is present.

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Case presentation

A female Caucasian baby was born by cesarean section at 39 weeks of gestation to a 22-year-old gravida 1, para 1 mother. The birth weight was 3650 g. No family history was known of any congenital diseases. There was no evidence of exposure to teratogens during pregnancy nor any history of consanguineous marriage. Casesarean section was performed because of acute life-threatening symptoms (centralization of blood circulation and circulatory failure were present) and generalized edema of the fetus diagnosed previously during prenatal ultrasound examination. Partial ectopia cordis and omphalocoele were not detected prenatally due to fetus generalized edema which prevented full visualization of both thoracic and abdominal wall.

Apgar score was 1, 6 and 9 at 1st, 3rd and 5th minute respectively. Resuscitation was established with Neopuff device immediately after delivery. pH obtained from umbilical cord blood was normal: 7.35 (BE – 0.1 mmol/L) and 7.34 (BE – 1.8 mmol/L).

After birth, physical examination revealed upper abdominal wall defect in the midline with omphalocoele and pulsative mass situated above omphalocoele. This was apex cordis located outside the thoracic cavity with no pericardium protection – recognized as a partial, thoracoabdominal type of ectopia cordis (EC) – a severe condition, sometimes associated with PC. Both of which are shown below (Fig. 1). A diagnosis of an incomplete pentalogy of Cantrell was established.

General edema and ascites were present, therefore peritoneal cavity puncture was done immediately after birth with decompression of 80 ml of exudative fluid. It is believed that fetal edema was due to cardiac failure.

The patient was admitted to Neonatal Intensive Care Unit, where additional volume of 270 ml of fluid was evacuated from peritoneal cavity within next five hours. Drainage of peritoneal cavity was maintained for the next 13 days.

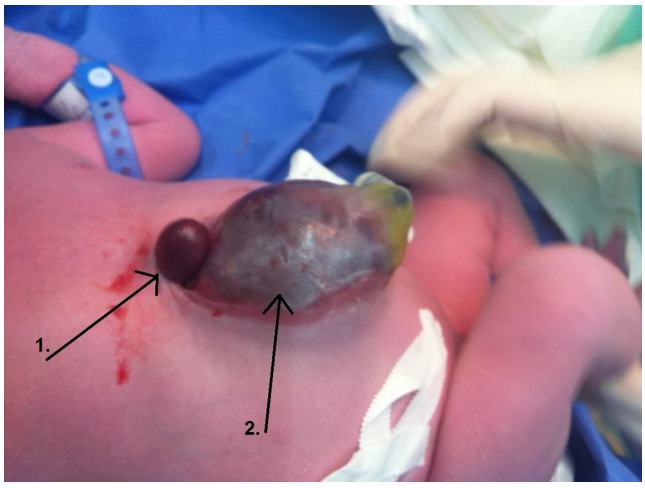

Fig. 1 – The picture was taken immediately after the birth. Arrow 1: Indicates partial ectopia cordis; Arrow 2: Indicates upper abdominal wall defect with omphalocoele

At 9th hour of life, respiratory difficulties appeared with tachypnea, grunting and retractions – the baby was supported with non-invasive ventilation and FiO_2 25%. Heart rhythm and blood pressure were found to be normal. Diagnostic approach to rule out infection was performed (CRP, complete blood count, blood smear and blood culture). Because chest X-ray showed inflammatory changes and CRP was above the normal range – antibiotic therapy was introduced. Blood culture was negative. The chromosome study showed a normal female karyotype (46,XX).

The consulting cardiologist found in echocardiographic study (ECHO) as follows: ventricular septal defect (1.0 cm) and foramen ovale (0.35 cm). The baby underwent surgery on 2nd day of life (DOL), that was a simultaneous operation including apex cordis relocation into thoracic cavity, pericardium closure and plastic surgery of omphalocoele and upper abdominal wall. During surgery baby was intubated and ventilated with SIMV mode. The postoperative treatment was not complicated with gradual respiratory improvement and extubation occurred on 6th DOL. The follow up ECHO study showed (apart from VSD) severe tricuspid insufficiency and severe mitral insufficiency. Diuretic therapy was started and continued (spironolacton and furosemide). Baby was on total parenteral nutrition for the first 12 days of life and initial trophic feeding was started on 13th DOL. At 18th DOL total enteral feeding was achieved. The patient was discharged from the hospital on 35 DOL.

The first cardiac follow up visit was at the end of 2nd month of age. Echocardiography study revealed perimembranous ventricular septal defect with bidirectional shunt, moderate pulmonary regurgitation, moderate mitral regurgitation and severe tricuspid regurgitation, the systolic pressure in the right ventricle calculated from tricuspid regurgitation was 100 mmHg. Due to the suspicion of pulmonary hypertension cardiac catheterization was performed. The study had a mean pulmonary arterial pressure 28 mmHg, Qp:Qs = 3.57, PVRI = 2.69 WU/m^2. The child was qualified to radical repair of congenital heart disease and 2 weeks later underwent cardiac surgery during which ventricular septal defect was closed with Dacron patch. The postoperative course was complicated by respiratory failure with pulmonary hypertension. There for sternum closing was postponed and the child required inhaled nitric oxide supply for 4 weeks. 8 weeks after surgery tracheostomy was carried out due to persistent respiratory failure. 9 weeks after cardiac surgery the child was removed from mechanical ventilation.

Cardiology follow up visits in the first year of life showed a small volume of the right ventricle, small atrial septal defect with right-to-left shunt, moderate tricuspid regurgitation and systolic blood pressure in the right ventricle calculated from tricuspid regurgitation 27–33 mmHg. Originally Glenn operation was planned. However, due to the gradual improvement of the child's cardiovascular endurance and increase of the right ventricle's volume, this surgery was not performed. Currently, at the age of 19 months, the child is waiting for surgery to close tracheostomy.