The complete spectrum of pentalogy of Cantrell in one of a set of dizygotic twins
A case report of a rare congenital anomaly

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Abstract

Rationale: Pentalogy of Cantrell (POC) is an extremely rare syndrome with an estimated incidence of 1:65,000 to 200,000 live births. Its complete form includes a midline epigastric abdominal wall defect, defects affecting the lower sternum, anterior diaphragm, diaphragmatic pericardium, and various intracardiac defects.

Patient concerns: We report a case of complete POC affecting only the first-born of a set of premature dizygotic twins.

Diagnosis: A giant omphalocele with an eviscerated liver and bowel on prenatal, obstetric ultrasonography at 24 gestational weeks was observed. At birth, physical examination confirmed a massive (10 × 8 cm) epigastric omphalocele in which a significant part of the liver was seen. A postnatal echocardiogram revealed the presence of an ostium secundum atrial septal defect, perimembranous ventricular septal defect, and moderate pulmonary stenosis. X-ray showed an abnormal intrathoracic positioned stomach, which was confirmed with a plain x-ray of the upper intestinal tract with hydrosoluble contrast. Computed tomography (CT) scan revealed the sternum’s absence and a close connection between the pericardial sac and the stomach wall.

Interventions: The patient underwent surgical intervention at 18 days of age.

Outcomes: Despite adequate and appropriate postoperative treatment, the baby rapidly deteriorated and died 72 hours after surgery.

Lessons: POC is a complex, high-mortality syndrome whose management requires a multidisciplinary approach and meticulous planning. Despite all efforts, POC carries a poor prognosis, particularly in patients affected by its complete form.

Abbreviations: ASD = atrial septal defect, CS = cesarian section, CT = computed tomography, EC = ectopia cordis, NICU = neonatal intensive care unit, POC = pentalogy of Cantrell, US = ultrasonography, VSD = ventricular septal defect.

Keywords: congenital anomalies, diagnosis, pentalogy of Cantrell, treatment, twins

1. Introduction

Pentalogy of Cantrell (POC) is an extremely rare syndrome with an estimated incidence of 1:65,000 to 200,000 live births, affecting men and women at a ratio of 1.35:1. Some 250 cases have been reported so far in the literature, mostly in the western countries (the United States and Europe). POC is a constellation of five congenital defects, including a midline epigastric abdominal wall defect, defects affecting the lower sternum, anterior diaphragm, diaphragmatic pericardium, and various intracardiac defects. However, the hallmark of POC is an omphalocele associated with ectopia cordis (EC). In some cases, the spectrum of the POC had not been present in a complete manner. Based on that, Toyama described the probable syndrome variant that includes 4 defects, intracardiac and ventral abdominal wall abnormalities, and the incomplete syndrome with various combinations of defects present, but always with sternal abnormalities. POC is associated with a high mortality rate that increases in cases of complex heart defects, pulmonary hypoplasia, associated malformations, and the existence of a complete presentation of this congenital disorder.

Herein, we report an extremely rare case of complete POC affecting only the first-born of a set of premature dizygotic twins.

2. Case report

A 33-year-old gravida 1, para 0, abortus 0, was referred to the obstetrics service at 34 gestational weeks for elective cesarean
section (CS) due to the finding of a giant omphalocele with an
eviscerated liver and bowel on prenatal, obstetric ultrasonogra-
phy (US) at 24 gestational weeks in one of a set of dizygotic twins.
The second twin was ultrasonographically normal. The parents
were counseled regarding ultrasound findings. The woman was
also diagnosed with preclampsia and gestational diabetes
mellitus, and methyldopa (250mg orally twice a day) was
administered. The family history was negative for congenital
anomalies or genetic abnormalities. There was no history of
consanguinity.

At delivery, the affected male twin had a birth weight of 2.050g
with the Apgar score of 4/6 in the first and fifth minute,
respectively. The infant was found to be cyanotic and hypotonic
with low respiratory effort. His peripheral oxygen saturation was
90% via pulse oximetry, respiratory rate was 48 breaths per
minute, the heart rate was 158 bpm, and blood pressure was 64/
32 mm Hg. Basic blood tests were normal. Physical examination
confirmed a massive (10 x 8 cm) cephalic (epigastric) omphalo-
cle in which a significant part of the liver was observed (Fig. 1).
The site of the omphalocele was covered with a saline-soaked
gauze pad. Following admission to the neonatal intensive care
unit (NICU), the baby was placed on nasal prong oxygen at 2L/
minute, the heart rate was 158bpm, and blood pressure was 64/
90% via pulse oximetry, respiratory rate was 48 breaths per
minute, and low respiratory effort. His peripheral oxygen saturation was

With the Apgar score of 4/6 in the 5th minute, the surgery
was postponed until the patient’s relative stabilization was
achieved. The patient underwent surgical intervention at 18 days
of age. After dissection of the omphalocele sac from the skin
edges, the ectopic heart in the right upper abdominal cavity,
bilateral anterior diaphragmatic defect with herniation of the
transverse colon and the stomach into the thoracic cavity, and
centrally located eviscerated liver were noted (Fig. 3). After
reducing the transverse colon into the abdominal cavity, defects
in the diaphragmatic pericardium and the right and the left
hemidiaphragm were reconstructed and sutured. Due to the
abdominal wall defect (10 cm in diameter) and the liver’s central
position, primary fascial reconstruction was not an option. The
closure of the omphalocele defect was performed using a silo bag.

After the procedures, the patient was taken intubated to the
NICU. Despite adequate and appropriate ventilation and high
doses of dobutamine and noradrenaline, the patient developed
inadequate oxygenation, progressive hypotension, and oliguria.
An adequate increase in pressure was not achieved even with
increased adrenaline doses added to the infusion pump. The
condition progressed with severe anuria and persistent metabolic
acidosis. The baby rapidly deteriorated and died 72 hours after
surgery.

3. Discussion

POC is an extremely rare syndrome with a poor prognosis.
POC includes 5 congenital defects, namely, a midline epigastric
abdominal wall defect, defects affecting the lower sternum,
intracardiac and ventral abdominal wall abnormalities); and
anterior diaphragm, diaphragmatic pericardium, and various
intracardiac defects. The hallmark of POC is an omphalocele
associated with EC. Toyama also described the probable
(incomplete) syndrome variant that includes 4 defects.[4] Our
study represents the first reported case of one of a set of dizygotic
twins with the complete POC in Bosnia and Herzegovina. The
presence of this complex anomaly in twins is extremely unusual,
but individual case reports have been published.[7,8] The etiology
of POC is still unknown and is considered to be of a
heterogeneous origin. It is believed that the lack of fusion in
the medial line of the mesoderm is responsible for the
development of this complex anomaly.[9] It usually occurs
sporadically, but familial cases have been reported indicating the
potential role of genetic factors in the pathogenesis of POC.[10]
Since no etiological factor has been identified in our case, we can
only hypothesize that hereditary and environmental factors,
individually or combined, could be responsible for the occurrence
of this anomaly.

Based on the number of malformations present, POC is
classified as complete or incomplete. Class I or certain diagnosis
implies a complete syndrome with all 5 defects present; Class II is
a probable diagnosis, with 4 defects present (including
intracardiac and ventral abdominal wall abnormalities); and
class III is an incomplete syndrome, with various combinations of
defects present, but always including a sternal abnormality.[3]
Accordingly, our patient fits class I or complete POC because all 5
defects were present.

POC abdominal wall defects can be presented as omphalocele,
epigastric hernia, umbilical hernia, diastasis recti, and combined
defects.[11] The most common abdominal wall defect is
omphalocele. Our patients had a giant cephalic (epigastric)
omphalocele in which the liver was centrally and mostly
eviscerated and located. Abnormalities affecting the sternum
can range from the short sternum, bifid sternum, absent xiphostom,
and defective lower part of the sternum to the sternum’s complete
absence. In our case, the sternum was absent with only a small
sternal ossification center. Defects in the heart pericardium,
mostly in the diaphragmatic pericardium, are integral compo-
nents of the full spectrum of POC syndrome and the anterior
diaphragmatic hernia, which in our case was bilateral. Failed
differentiation of the mesoderm into the septum transversum
during early fetal development is considered the reason for the
emergence of POC components. Various heart defects have been
reported in infants with POC, including VSD, ASD, pulmonary stenosis, dextrocardia, and tetralogy of Fallot. The most severe cardiac anomaly in POC patients is EC. Our patient also had EC with the heart located within the abdominal cavity, to the right of the medial line, below the omphalocele sac. The findings that VSD (100%) and ASD (53%) are the most common intracardiac anomalies in POC were also confirmed in our case.[3]

POC management represents a tremendous multidisciplinary challenge involving pediatric surgery, cardiovascular surgery, anesthesiology, and NICU.[12] Early surgical correction is the preferred treatment modality to minimize the risk of heart complications and reduce fluid loss and infection onset due to uncorrected omphalocele.[13] However, early surgical intervention might be associated with an increased mortality rate.[13] The usefulness of initial conservative treatment in stable patients has also been shown by using antibiotic prophylaxis and dressing with topical antimicrobial agents of the omphalocele sac until its epithelialization. In the present case, based on the multidisciplinary approach, delayed surgery was planned and performed.

Postoperative intensive care is essential to prevent or reduce hypoxia-related complications caused by pulmonary hypoplasia, surgical corrections of congenital malformations, and increased intra-abdominal pressure after the closure of the anterior abdominal wall.

POC is considered a high-mortality syndrome. Survival is related to the severity and complexity of heart malformations and EC location.[14] Late mortality is caused by complications of heart dysfunction, infections, or adhesive small bowel obstruction.[14,15] Due to the patient’s preoperative unstable status and the heart’s postoperative inability to adjust to the intrathoracic conditions, our patient died shortly after surgery.

In conclusion, POC is a high-mortality syndrome with a poor prognosis, particularly in patients affected by the POC complete form. Timely identification of all aspects of this syndrome,
supportive care, and the choice of an optimal surgery modality (primary/staged) are the unique challenges of a multidisciplinary team in managing patients with POC.

Author contributions

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