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REVIEW ARTICLE

A LOCAL CASE OF PENTALOGY OF CANTRELL IN 2013 (CLINICAL CASE STUDY)

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ABSTRACT

Qualified as a very rare disease, the Pentalogy of Cantrell comprises a number of defects, such as: anterior abdominal wall defect, lower sternum defect, diaphragmatic and pericardial defects, and congenital heart malformations. The typical form of the pentalogy includes all these five defects. The cases of the complete pentalogy described in the existing literature are only a few. The first description of the pentalogy was made by Cantrell et al. in 1958, who gave an account of 5 cases presented with this abnormality. Cases with only two, three or four of the defects are more frequent. The case we present here involves the 16-year-old A.M.A., pregnant in the 18th gestation week. The fetus is male with bilateral cleavage of lip and palate; low-set ears, congenitally deformed feet (pes equinovarus), absent little toe of the right foot, complete absence of sternum and whole abdominal wall defect. The heart, liver and intestinal loops, as well as the kidneys and urinary bladder are ectopically located outside their corresponding cavities. A lack of pericardium is noted, together with ventricular septal defect, and atrial septal defect. My colleagues and I believe this case is typical for Pentalogy of Cantrell.

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INTRODUCTION

A.M.A., age 16, pregnant in 18th gestation week, of Roma ethnic origin. Admitted to the university clinic with a suspected abortion, and frequent painful contractions. The patient hasn't visited women's consultation centre regularly. No screening has been performed in the first trimester. In the hospital, during the echographic study, I found the heart was outside the chest cavity and the liver, the kidneys and intestines were outside the abdominal cavity (Pictures 1, 2, 3). Furthermore, the echograph study showed bilateral cleavage of the palate and lip and congenitally deformed feet. An increased quantity of amniotic fluid; heart – turned upwards and to the left. The fetus was male. At the sight of the ectopical heart I suspected the Pentalogy of Cantrell. The presence of the remaining structural defects gave enough evidence for genetic study. There were no chromosome abnormalities. The study showed a normal male cryotype. (Skin biopsy after discarding the fetus.) Because of the found abnormalities, which revealed an impossible life after birth, as well as due to the minor age of the pregnant, we contacted her relatives. It was decided that the pregnancy should be terminated. After birth, the fetus did not show any vital signs, apart from occasional heartbeat. During the post mortem we found out cleavage of the lip and palate, low-set ears, congenitally deformed feet (pes equinovarus), absent little toe of the right foot, complete absence of sternum and whole abdominal wall defect. Absence of pericardium, ventricular septal defect, and atrial septal defect were found as well. (Pictures 4,5,6,7) Prenatal diagnosis of Pentalogy of Cantrell could have been made even during the first trimester of pregnancy. Differential diagnosis then could have been made with omphalocoele, which is observed physiologically up to the 10-12 gestation week. However, if in the sac, apart from the intestinal loops, other organs can be seen, especially if this is ectopically situated heart, we will suspect Pentalogy of Cantrell. The prognosis for the Pentalogy of Cantrell depends on the severity of the internal and additional heart defects, pulmonary hypoplasia, the degree of abdominal wall defect, brain abnormalities and diaphragmatic hernia. The average survival period without operation is about 36 hours. Studies have shown that even with special care in professional centres and multiple correction operations, mortality still remains high.

DISCUSSION

The case presented here has all defects typical for Pentalogy of Cantrell, including bilateral deep cleavage of lip/palate and congenitally deformed feet (equinovarus). Pentalogy of Cantrell is characterized by a combination of congenital defects.

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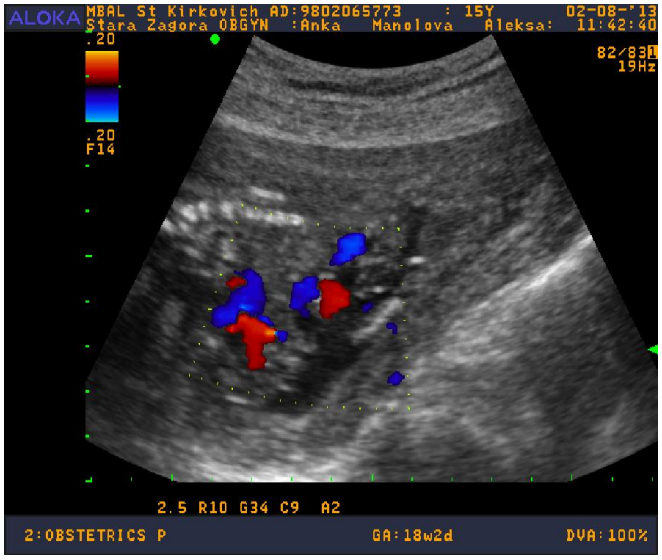
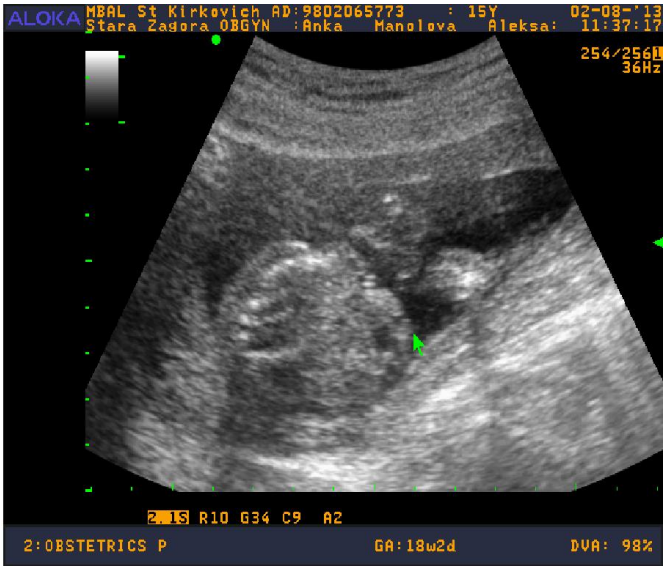
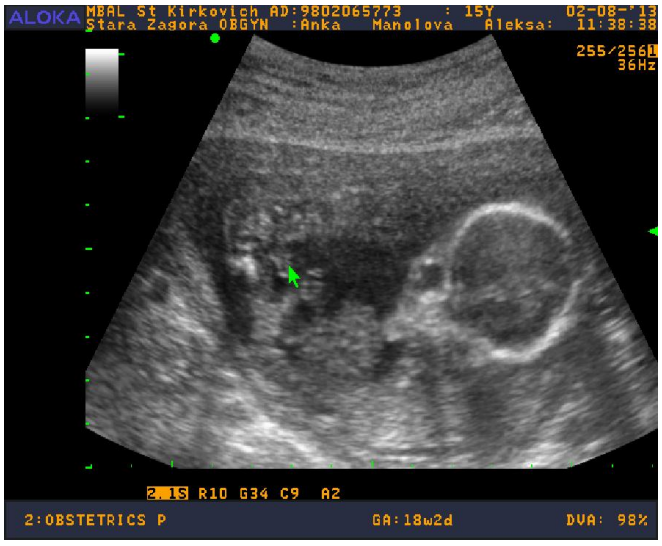


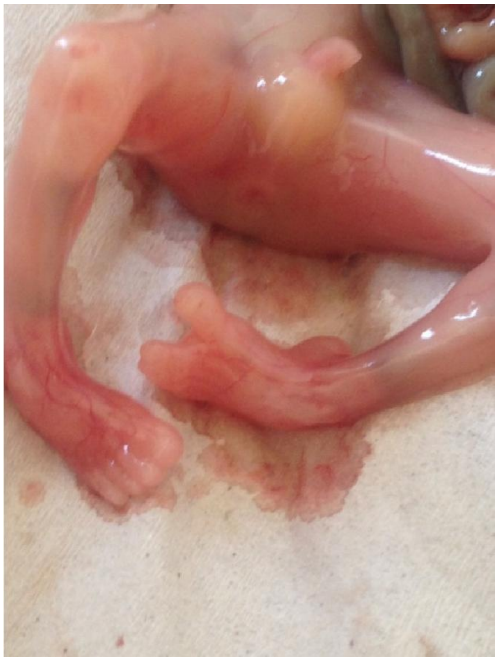
Picture 1. Heart outside chest



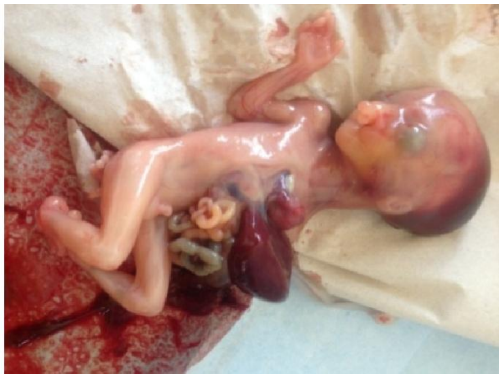
Picture 2. Liver outside the abdominal cavity



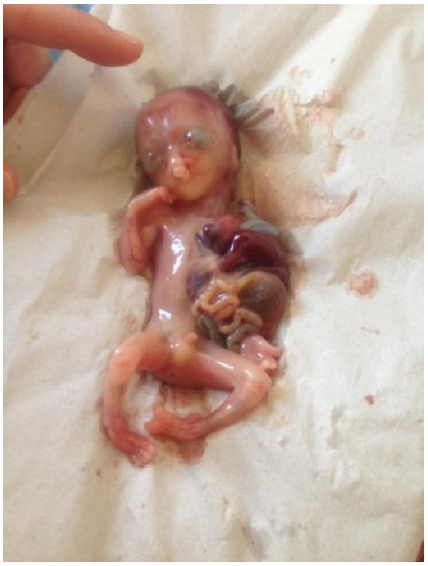
Picture 3. Abdominal organs outside the abdomen



Picture 4. Pes equinovarus end lack of a finger

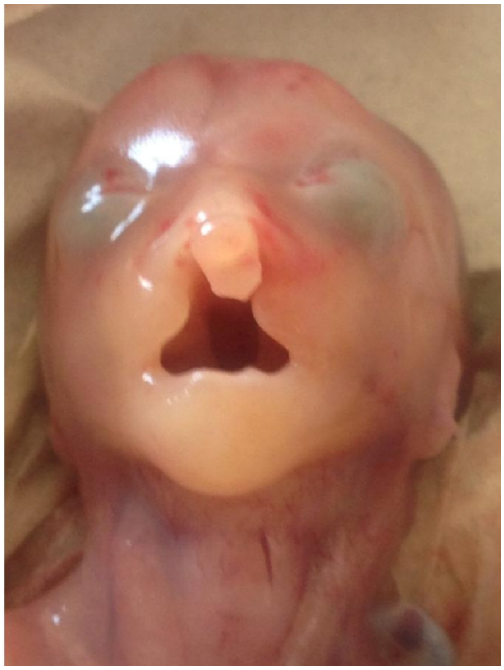


Picture 5.



Picture 6.

Picture 5. 6. Heart and abdominal organs outside the abdominal and thoracic cage



Picture 7.

1. Anterior abdominal wall defect– omphalocoele
2. Defective or completely absent sternum – Ectopically located heart (outside the chest cavity) – ectopia cordis
3. Defective or absent diaphragm (diaphragmatic defect)
4. Pericardial defect.
5. Cardiac malformations of the fetus

Ventricular septal defect (VSD), atrial septal defect (ASD), tetralogy of Fallot, left ventricular diverticulum. The etiology of the Pentalogy is unclear. Most cases are sporadic and have not been recorded. There is a 2,7:1 male to female ratio. Due to different phenotypes of abdominal wall defects in the Pentalogy of Cantrell, it is said that the disease is also caused by multiple factors, including mechanical, teratogenic causes, gene mutations, chromosome abnormalities, such as trisomia 13 and 18, etc. During this syndrome other abnormalities have been described, such as craniofacial abnormalities – cleavage of the lip and/or the palate; abnormalities of the central nervous system, such as hydrocephalus; skeletal malformations such as equinovarus. The sternum defects include bifurcated sternum (26%), a lack of xiphoid process (10%), and absence of the lower two-thirds of the sternum (9%). The anterior abdominal wall defect is most often omphalocoele (63%). The ventral retrosternal defect of the diaphragm is observed in 91% of all cases. With prenatal echography, the Pentalogy of Cantrell can normally be diagnosed as early as in the 10^{th} gestation week. It is suggested that MRI and prenatal fetal echocardiography provide optimal evaluation of fetuses with this syndrome. The use of 2D ultrasound in the first trimester, and the accompanying use of 3D ultrasound can assist the improvement of the visualization of fetal abnormalities in different rectangular planes, even with unfavorable fetal positions. If the diagnosis is based on an ultrasound study, chromosome analysis for trisomes 13 and 18, and Turner's syndrome is recommended.

REFERENCES

Cantrell JR, Haller JA, Ravith MM. 1958. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and heart. *Surg Gynecol Obstet*., 107:602–614.

Chen CP. 2007. Syndromes and disorders associated with omphalocele (II): OEIS complex and Pentalogy of Cantrell. *Taiwan J Obstet Gynecol*., 46(2):103–10.

Forzano F, Daubeney PE, White SM. Midline raphé, sternal cleft, and other midline abnormalities: a new dominant syndrome? *Am J Med Genet A*.

O'Gorman CS, Tortoriello TA, McMahon CJ. 2009. Outcome of children with pentalogy of Cantrell following cardiac surgery. *Pediatr Cardiol*, 30(4):426–30.

Van Allen MI, Curry C, Walden CE, Gallagher L, Patten RM. 1987. Limb-body wall complex: II.Limb and spine defects. *Am J Med Genet*, 28(3):549–65.

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