

Prenatal Diagnosis of Pentalogy of Cantrell in the Third Trimester

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Abstract: We report about the prenatal diagnosis of Pentalogy of Cantrell in the third trimester. In this case sonographic evaluation revealed mild form of ectopia cordis, severe omphalocele, small meningomyelocele, mild hydrocephalus, severe polyhydramnios and allantoic cyst of the umbilical cord. The pregnancy was terminated by a caesarean section at 35 weeks of pregnancy because of severe polyhydramnios. The neonate died shortly after delivery and the obduction confirmed the diagnosis of Pentalogy of Cantrell. We discuss the reason of the late prenatal diagnosis in this case, the importance of early prenatal diagnosis and the options of pregnancy management.

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Introduction

Pentalogy of Cantrell, first described by Cantrell in 1958 by five neonates [1], is a very rare congenital syndrome consisting of abdominal wall defect, ectopia cordis, lower sternal defect, pericardial and anterior diaphragmatic defect. This syndrome has several synonyms: Cantrell-Heller-Ravitch syndrome, thoraco-abdominal ectopia cordis, pentalogy syndrome or peritoneopericardial diaphragmatic hernia. The expression of these defects is variable and therefore much more incomplete expressions have been reported. The prevalence of Pentalogy of Cantrell (POC) is very rare and it ranges from 1:65 000 to 1:200 000 births [2]. Less than 100 cases have been reported in the literature. Aetiology of POC is unknown, but is more often associated with aneuploidies [3, 4]. The pathogenesis of Pentalogy of Cantrell is associated with a failure of the lateral mesodermal folds to migrate to the midline, causing the abdominal defects, and failure of the septum transversum to develop, causing defects in the anterior diaphragm and pericardium between 14 and 18 days after conception. There is no evidence of recurrence risk, but one case of monozygotic twins concordant for the syndrome has been reported [5]. Associated anomalies include cranial and facial anomalies, intracardiac anomalies, aneuploidies, cystic hygroma, dysplastic kidneys, clubfoot, hydrocephalus, and anencephaly. Differential diagnosis includes isolated ectopia cordis by amniotic band syndrome, isolated omphalocele or gastroschisis, Beckwith-Wiedemann syndrome and body stalk anomaly. The prognosis depends primarily on the severity of ectopia cordis, on the size of the abdominal wall defect and presence of associated anomalies. The abdominal wall defect should be repaired immediately after delivery by using a mesh, but the surgical treatment is difficult because the infant's abdomen is abnormally small because it had no need to expand to accommodate the developing organs.

We report the prenatal diagnosis of Pentalogy of Cantrell in the third trimester and we discuss the importance of early prenatal diagnose.

Case report

In April 2008, a 29-year-old Caucasian primigravida was referred to our department at 29 weeks of her pregnancy, with a suspicion of omphalocele and severe polyhydramnios.

Both the husband and the wife are Slovaks, and neither has a family history of a hereditary disorder. The mother did not take any drug in the first three month of pregnancy and also not before conception. Exposure to other teratogens was negative. Her previous examinations were performed in England, where she conceived and spent her 28 weeks of pregnancy. All results were reported to be normal. First trimester screening and triple test were not performed. The ultrasound examination in 10th week and 20th week of her pregnancy were reported to be normal.

We performed level II ultrasound scan in our clinic. Transabdominal ultrasound examination using an DC-6 ultrasound system (Shenzen Mindray Bio-Medical Electronics) with a multi-frequency transducer revealed vital monofetal pregnancy with a composite sonographic age of 31 weeks + 0 days, mild form of ectopia cordis, severe omphalocele, small meningomyelocele, mild hydrocephalus, severe polyhydramnios, allantoic cyst of the umbilical cord. We found a large ventral wall defect with extruded liver, stomach, bowel through the wall defect (Figure 1). The extent of the ectopia cordis was small, and no intracardiac abnormalities were detected (Figure 2). We used 3D rendering to obtain scans of the foetal face, and we didn't find any anomalies. Gender was female. Level III ultrasound scan confirmed our findings. Serological tests TORCH, HIV, HBsAg, RRR, TPHA were negative. According to the malformations found and complete clinical status, the



Figure 1 – Large ventral wall defect with extruded liver, stomach, bowel through the wall defect.



Figure 2 – Mild form of ectopia cordis.

diagnosis of Pentalogy of Cantrell was assessed. The parents were informed about the poor prognosis of foetus, but they refused karyotypisation. They preferred conservative management of pregnancy and refused also amniodrainage. The foetus was monitored by ultrasound in one week intervals. In the 35th week of gestation polyhydramnios caused, severe abdominal pain, due to the distension of the uterus. The pregnancy was terminated by a caesarean section at 35 weeks of pregnancy because of marked polyhydramnios. The neonate died of respiratory insufficiency 60 minutes after delivery and the obduction confirmed the diagnosis of Pentalogy of Cantrell. Karyotype analysis revealed a normal 46, XX neonate.

Discussion

Pentalogy of Cantrell is a very rare congenital syndrome defined by supraumbilical wall defect, to date less than 100 cases have been reported in the literature. Larger patient files were described by a few of authors. In 1972 Toyama reviewed 60 postnatally diagnosed cases, and he reported a survival rate of 20% [6]. Ghidini reported in 1988 of 17 prenatally diagnosed cases, with a survival rate of 0% [7]. The extents of defects in prenatally diagnosed cases were to notable to be detected, and these cases could have been much more severe than postnatally diagnosed cases. But the prognosis is determined by many others anomalies associated with Pentalogy of Canrell [8]. Pentalogy of Cantrell, which is characterized by two major defects: ectopia cordis and an abdominal wall defect should be detected in the first trimester. In 1991 Bennett et al. reported as the first about the prenatal diagnosis of Pentalogy of Cantrell in the first trimester [9]. It is important to allow the pregnant woman to make a decision even before first foetal movements. Tools like 3D sonography can help during the process of prenatal counselling. The 3D visualization allows better understanding of the anatomic conditions [10].



Figure 3 – Allantoic cyst of the umbilical cord.

We presented a case of Pentalogy of Cantrell diagnosed in the third trimester. Unfortunately, ultrasound examinations performed in England were reported to be normal. Neither first trimester screening, nor triple-test was performed. Simple alpha-fetoprotein estimation, usually routinely performed there, where first trimester combined test is not available, should be of diagnostic value in this case [11]. We conclude, that first trimester ultrasound screening usually performed in the 12th–13th week of pregnancy would allow making the correct diagnosis in this case.

The foetus presented herein, exhibits one interesting anomaly, allantoic umbilical cord cyst (Figure 3), which is often associated with omphalocele [12].

In our case was the pregnancy terminated by a caesarean section at 35 weeks of pregnancy because of maternal pain due to marked polyhydramnios. However, if amniodrainage could have been performed, spontaneous delivery at term would have been preferable. Studies addressing this interrogation failed to show a difference in foetal morbidity and mortality between vaginal delivery and caesarean section [13, 14, 15, 16, 17, 18].

Management of a severely malformed foetus with poor prognosis in the second and trimester of pregnancy depends on several factors. First factor is the attitude of the mother. Second are the law conditions of the particular country. The Slovakian law does not allow terminating pregnancy later than in 24 weeks of pregnancy. And in this case, the mother insisted on conservative management of the pregnancy. If it were legal, the option of pregnancy termination at any stage of pregnancy would be method of choice.

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