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First Trimester Diagnosis of Pentalogy of Cantrell: A Case Report

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Abstract

Objective: Pentalogy of Cantrell is a very rare congenital malformation complex, in which the intrathoracic and intraabdominal organs are completely or partially located outside the body secondary to the thoracoabdominal body wall defects.

Case: In this report, a 23-year-old primigravid woman who admitted to our perinatology clinic in the 12th week of her pregnancy was presented. Following the first trimester scanning, the fetus was diagnosed as having a large abdominal defect, an ectopia cordis and club foot. Informed consent was obtained and termination of pregnancy was performed.

Conclusion: Neonatal prognosis of the fetuses with pentalogy of Cantrell is generally poor and termination of pregnancy should be offered in cases prenatally diagnosed at early weeks.

Keywords: Pentalogy of Cantrell, ectopia cordis, first trimester.

İlk trimesterde tanısı koyulan Cantrell pentalojisi: Olgu sunumu

Amaç: Cantrell pentalojisi torakal ve abdominal defektlere eşlik eden, intratorasik-intraabdominal organ evisserasyonları ile karakterize, son derece nadir görülen bir anomali kompleksidir.

Olgu: Bu vaka sunumunda 23 yaşında primigravid, 12. gebelik haftasında ultrasonografi aracılığıyla Cantrell pentalojisi tanısı konulan bir olgu tanımlanmaktadır. Obstetrik ultrasonografisinde fetusta geniş omfalosel, ektopia kordis ve pes ekinovarus anomalileri saptanmıştır. Ailenin onayı ile gebelik terminasyonu uygulanmıştır.

Sonuç: Neonatal cerrahi sonuçları genelde kötüdür ve erken gebelik haftalarında tanı alan olgulara gebelik sonlandırması önerilmektedir.

Anahtar Sözcükler: Cantrell pentalojisi, ektopia kordis, ilk trimester.

Introduction

Pentology of Cantrell or thoracoabdominal ectopia cordis defined by Cantrell in 1958 for the first time is an anomaly complex characterized by sternal, anterior, diaphragmatical, pericardial or supraumbilical abdominal defects together with heart located out of thorax.[1] Its incidence is 100,000 and it is 0.5-0.7 in pregnancy, but its certain etiology is not known.[2]

Though prenatal diagnosis is easy, most of the cases can be diagnosed at second trimester. Cardiac anomalies such as atrial septal defect, ventricular septal defect and Fallot tetralogy frequently accompany to the syndrome and the most important factor determining the prognosis is the severity of intracardiac anomalies.[3] Rare cases which live by corrective surgery performed in neonatal period are reported in the literature.[4, 5]

In this case report, a case diagnosed as pentalogy of Cantrell by ultrasonography at 12th gestational week and the management are discussed in company with current literature information.

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Case Report

Twenty-two years old patient with Gravida 1 and Parity 0 applied to our clinic at her 12th gestational week for routine follow up. There was no prominent diagnosis in the obstetric history or family history of the patient. In the obstetric ultrasonography, single, living fetus with 58 mm crown-rump length and compatible with 12 weeks was detected. In the ultrasonographical examination of fetus, sternal defect, ectopia cordis and abdominal anterior wall defect were detected (Figure 1).

It was seen that liver and intestinal structures were herniated due to abdominal anterior wall defect. Flow in ductus venosus was evaluated as normal in the colored Doppler sonography. In the light of current findings, nuchal translucency (NT) value of the fetus diagnosed as pentalogy of Cantrell was measured as 2.7 mm (bigger than 95. percentile). Though any major anomaly was not detected fetal extremities, pes equinovarus deformity was detected in both lower extremities. Examination by 3D ultrasonography could not be performed. Current findings and the prognosis of the disease were told to the family. After their informed consent was taken, pregnancy was terminated within 9 hours by totally 800 microgram vaginal misoprostol. In the macroscopic examination of 34 gram female fetus, a wide median defect including abdominal anterior wall and thorax, and intraabdominal organs herniated by this defect and ectopic located heart were observed (Figure 2).

Skin biopsy was taken from the fetus for genetic examination. As a result of the genetic examination, it was found as 46, XX normal karyotype. In the pathological examination, thoracic ectopia cordis together with sternum defect, abdominal anterior wall defect and herniation of intraabdominal organs were observed in the fetus.

Discussion

Pentalogy of Cantrell is a quite rare anomaly in eviscerations of intrathoracic-intraabdominal organs accompanying thoracic and abdominal defects. All components of the syndrome rarely coexist. In the series of Toyoma et al.[6] including 61 cases, it is categorized into three groups according to the existence of components forming pentalogy. In type 1, all five defects exist diagnostically. In type 2, four or five defects are together with an estimated diagnosis. In type 3, only incomplete expression of defects exists in various degrees.[6]

Pathogenesis of the syndrome is not known exactly. Thoracic and abdominal wall develop-

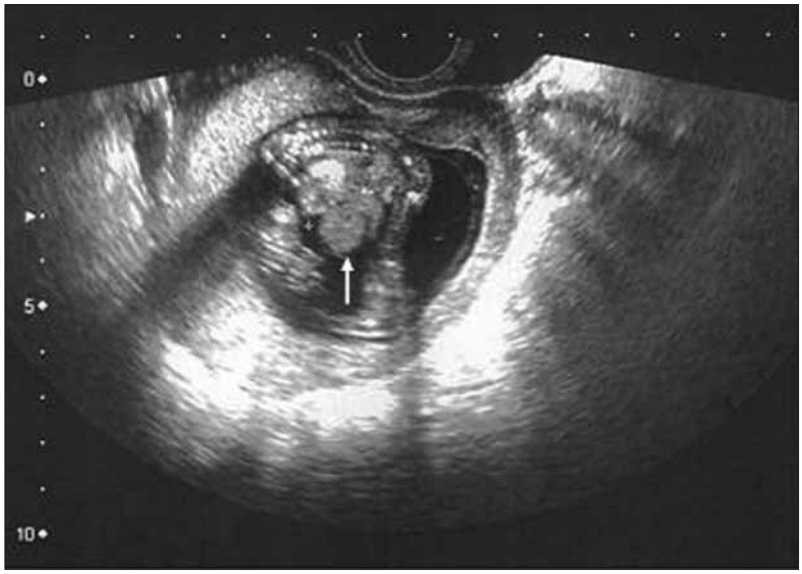

Figure 1. Appearance of abdominal anterior wall defect (white arrow) including liver tissue in first trimester and ectopic located heart (+ sign).

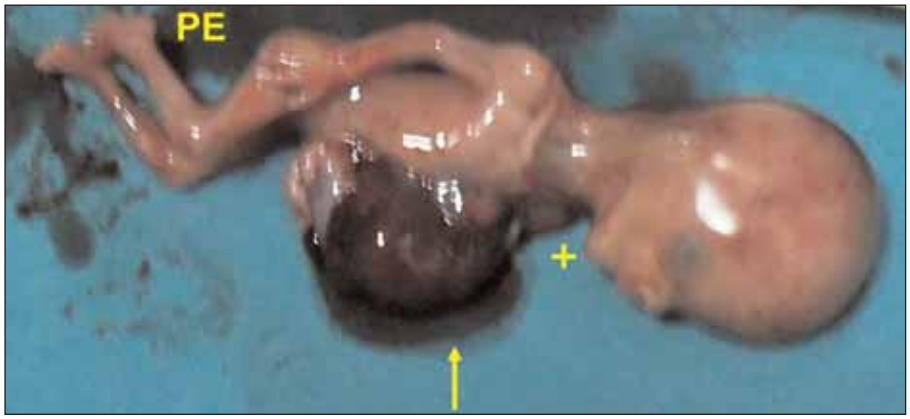

Figure 2. The appearance of ectopic cordis (+ sign), herniated liver and intestinal segments (white arrow) and 12 week fetus having pentalogy of Cantrell with pes equinovarus (PE).

ments are completed at 9th week of embryo. It is reported that if any defect appears on the midline fusion of embryonic lateral mesoderms due to genetic or external reasons at this period, clinical situations may appears varying from isolated ectopia cordis to complete ventral evisceration according to the location and severity of defect.[6,7]. Some researchers reported that pentalogy of Cantrell may occur by the effect of "mechanical teratogenity" caused by chorionic or yolk sac rupture.[8]

In complete cases, the diagnosis of the syndrome can be established easily beginning from 10th gestational week; however, especially as in our case, the diagnosis can be established on second trimester in 60% of cases with incomplete defects or isolated ectopia cordis .[9] Our case is an example in terms of establishing early diagnosis. Heart out of thorax together with the existence of liver and intestine segments eviscerated by abdominal anterior wall defect in ultrasonographic examination is diagnostic[7,10] In first trimester cases where it is suspicious to diagnose, 3D ultrasonography or fetal magnetic resonance examination may provide more detailed information about disease.[10]

Pentalogy of Cantrell is accepted as a sporadic malformation group, but also hydrocephaly, encephalocele, cloacal exstrophy and intrinsic cardiac defects may accompany to the syndrome.[4-6] In our case, bilateral club foot deformity was detected in addition to the pentalogy. Most frequently defined chromosomal anomaly associated with pentalogy is Trisomy 18 which can be found in 5-10% of cases.[1-4] Chromosome examination result of our case was found as 46, XX normal karyotype. Before giving consultancy about prognosis, prenatal diagnosis should certainly be suggested to couples who do not want the option of dispatch by current diagnosis.

The prognosis in pentalogy of Cantrell varies according to other accompanying anomalies and intrinsic heart anomalies; however, syndrome generally is accepted as lethal.[6-9]. Gestational termination is reported as a proper approach in early gestational weeks.[2-5] An atraumatic cesarean delivery and then a corrective surgery are recommended in cases that have isolated ectopic heart anomaly and normal karyotype. It was reported that rarely positive results are obtained by corrective surgery applied to these cases at neonatal period.[4,5] It was accepted that it is the most significant factor causing low surgical success of major vessel bendings caused by the reduction of cavities of visceral organs such as heart and liver.

Conclusion

Consequently, as a rare congenital anomaly group, pentalogy of Cantrell requires a proper perinatal evaluation plan when it is detected at prenatal period. Fetus should be investigated in terms of other accompanying anomalies and intrinsic cardiac defects. According to the literature, termination is suitable when anomaly is detected at

early gestational weeks or in case of the existence of accompanying chromosomal anomalies. Atraumatic cesarean delivery can be applied by pediatric surgery under consultation in cases with isolated ectoia cordis and with diploid chromosome diagnosed after 2nd trimester.

References

1. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. *Surg Gynecol Obstet* 1958;107:602–14.
2. Desselle C, Herve P, Toutain A, Lardy H, Sembely C, Perrotin F. Pentalogy of Cantrell: sonographic assessment. *J Clin Ultrasound* 2007;35:216-20.
3. Hornberger LK, Colan SD, Lock JE, Wessel DL, Mayer JE. Outcome of patients with ectopia cordis and significant intracardiac defects. *Circulation* 1996;94:32-7.
4. Korver AM, Haas F, Freund MW, Strengers JL. Pentalogy of Cantrell: successful early correction. *Pediatr Cardiol* 2008;29:146-9.
5. Samir K, Ghez O, Metras D, Kreitmann B. Ectopia cordis, a successful single stage thoracoabdominal repair. *Interact Cardiovasc Thorac Surg* 2003;2:611-3.
6. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium and heart: a case report and review of the syndrome. *Pediatrics* 1972;50:778–92.
7. Önderoğlu L, Baykal C, Tulunay G, Talim B, Kale G. Prenatal diagnosis of Cantrell's pentalogy: a case report. *Turk J Pediatr* 2003;45:357-8.
8. Groner JI, Ziegler MM, Azizkhan RG, Weber TR. Operative Pediatric Surgery. New York: McGraw-Hill Inc, 2003;279-93.
9. Repondek-Liberska M, Janiak K, Wloch A. Fetal echocardiography in ectopia cordis. *Pediatr Cardiol* 2000; 21:249-52.
10. Peixoto-Filho FM, Do Cima LC, Nakamura-Pereira M. Prenatal diagnosis of pentalogy of Cantrell in the first trimester: is 3-dimensional sonography needed? *J Clin Ultrasound* 2009;37:112-4.
11. McMahon CJ, Taylor MD, Cassady CI, Olutoye OO, Bezold LI. Diagnosis of pentalogy of Cantrell in the fetus using magnetic resonance imaging and ultrasound. *Pediatr Cardiol* 2007;28:172-5.
12. Murata S, Nakata M, Sumie M, Mastsuabra M, Sugino N. Prenatal diagnosis of pentalogy of Cantrell with craniorachischisis by three-dimensional ultrasonography in the first trimester. *Taiwan J Obstet Gynecol* 2009;48: 317-8.
13. Gün I, Kurdoğlu M, Müngen E, Muhcu M, Babacan A, Atay V. Prenatal diagnosis of vertebral deformities associated with pentalogy of cantrell: the role of threedimensional sonography? *J Clin Ultrasound* 2010;38:446-9.