Prenatal diagnosis of Cantrell's pentalogy: a case report

Lütfü Önderoğlu[1], Cem Baykal[2], Gökhan Tulunay[2], Beril Talim[3], Gülsev Kale[3]

Departments of [1]Obstetrics and Gynecology, and [3]Pediatrics, Hacettepe University Faculty of Medicine and [2]Social Security Ankara Maternity Hospital, Ankara, Turkey

SUMMARY: Önderoğlu L, Baykal C, Tulunay G, Talim B, Kale G. Prenatal diagnosis of Cantrell's pentalogy: a case report. Turk J Pediatr 2003; 45: 357-358.

A 23 year-old nulliparous woman was admitted to the obstetrics clinic in the 12th week of her pregnancy. Following the first trimester scanning, the fetus was diagnosed as having a large omphalocele and ectopia cordis. It was thought to be a thoracoabdominal wall defect and a possible case of Cantrell's pentalogy. Amniocentesis was performed and at the 16th week, the pregnancy was terminated because of karyotype revealing trisomy 21 and the serious structural defects. Autopsy demonstrated an ectopia cordis without pericardium and an abdominal wall defect with an omphalocele. Fetus had no diaphragma or sternum, and pulmonary and extremity anomalies were also present. With these findings, this case is suggested to be a variant of Cantrell's pentalogy.

Key words: Cantrell's pentalogy, trisomy 21 prenatal diagnosis.

Isolated ectopia cordis is defined as an anomaly in which the fetal heart lies outside the thoracic cavity; it is accepted as a poor prognosis anomaly[1]. Despite successful surgical correction, only a few cases have been reported to survive. Ectopia cordis is also a component of Cantrell's pentalogy, of which other components are abdominal wall defects with omphalocele, and sternum, diaphragm and congenital intracardiac anomalies. It is a very rare anomaly with an incidence of less than 1/100,000. Until recent decades, prenatal diagnosis was very difficult. Now it is possible to diagnose almost all ectopia cordis and coexisting anomalies in the first trimester. We present Cantrell's pentalogy which, as far as we know, is the first case associated with trisomy 21.

Case Report

A 23-year-old nulliparous woman was admitted to Hacettepe University Hospital's obstetrics clinic in the 12th week of her pregnancy. Parents were first-degree relatives, and there was no history of anomalous child in their families. First trimester scanning was performed and a living fetus with regular heartbeat was seen, but the fetus had a large omphalocele, which contained the liver and intestines. More interesting, the fetal heart was seen over the omphalocele and clearly out of the thorax (Fig. 1). Poor prognosis of the fetus was explained to the family and a decision for close follow-up was taken. An amniocentesis was performed thereafter and fetal karyotype revealed trisomy 21. Pregnancy was terminated at the 16th week because of trisomy 21 and structural defects.

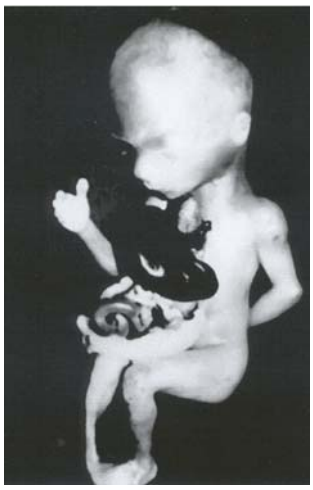

Fig. 1. Sonographically detected anomalies of 12-week fetus: heart is seen out of the thorax over the omphalocele, which contains the liver and intestines.

A complete autopsy was performed. It was a 125 g female fetus, with a crown-rump length of 12 cm and heel toe length of 2.4 cm. In the midline, just at the right side of the normally placed umbilicus, a 3 cm long ventral abdominal wall defect extending cranially towards the thoracic wall was present. From this defect, the heart, liver, stomach, intestines and spleen were all placed outside the thoracoabdominal cavity (Fig. 2). The heart showed a configuration of cephalic pointing cardiac apex, and pericardium was completely absent; no other cardiac malformations were detected. The diaphragm was identified only as a remnant at the posterior thoracic wall; complete absence is perhaps more accurate. There was no evidence of the sternum. An incomplete fissure of the right lung was present and the liver was enormously large compared with other organs. Additionally, bilateral simian lines, flexion contracture of the 3rd finger of the right hand and talipes equinovarus deformity of the right foot were present. No other pathological findings were documented in other organ systems. The fetus was reported as having trisomy 21 by karyotype analysis; karyotypes of the parents were normal. It was thus concluded that this is a case of Cantrell's pentalogy combined with trisomy 21.

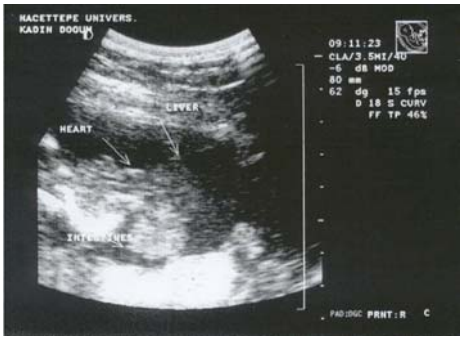

Fig. 2. Gross view of fetus showing heart, liver, and intestines located outside the thoracoabdominal cavity, talipes equinovarus of the right foot and flexion contracture of the right 3rd finger.

Discussion

Prenatal diagnosis of ectopia cordis was reported by Sepulveda et al.[2] and Bennett et al.[3] at 12 weeks and 11 weeks of gestation respectively. Liang et al.[4] recently reported a case of Cantrell's pentalogy at 10 weeks' gestation. In this case, correct diagnosis of ectopia cordis and omphalocele were possible with ultrasonography at the 12th week of gestation.

They key features of the case were a thoracoabdominal ectopia cordis, a large omphalocele (containing liver, spleen, stomach and intestines) also and absence of pericardium, sternum and diaphragm. Minor extremity anomalies were also present. Absence of sternum and diaphragm are anomalies that are very difficult to demonstrate by ultrasonography at such an early gestational age. Another component of Cantrell's pentalogy is congenital intracardiac defects, but it is also very difficult to demonstrate this defect at early gestational weeks.

In this case, the combined defect of ectopia cordis and omphalocele suggested Cantrell's pentalogy and autopsy findings confirmed the diagnosis. There are a few cases of isolated ectopia cordis associated with chromosomal anomalies and trisomies[5] in the literature, but Cantrell's pentalogy with trisomy 21 has not been reported before, to our knowledge. Because of the poor prognosis of Cantrell's pentalogy, early antenatal sonographic detection of it is important and allows for elective abortion before viability.

REFERENCES

1. Knox L, Tuggle D, Knott-Craig CJ. Repair of congenital sternal clefts in adolescence and infancy. J Pediatr Surg 1994; 29: 1513-1516.
2. Sepulveda W, Weiner E, Bower S, Flack NJ, Bennett PR, Fisk NN. Ectopia cordis in atriploid fetus: first-trimester diagnosis using transvaginal color Doppler ultarosongraphy and chorionic villus sampling. J Clin Ultrasound 1994; 22: 573-575.
3. Bennett TL, Burlbaw J, Drake CK, Finley BE. Diagnosis of ectopia cordis at 12 weeks gestation using transabdomina ultrasonography with color flow Doppler. J Ultrasound Med 1991; 10: 695-696.
4. Liang RI, Huang SE, Chang FM. Prenatal diagnosis of ectopia cordis at 10 weeks of gestation using two-dimensional and three-dimensional ultrasonography. Ultrasound Obstet Gynecol 1997; 10: 137-139.
5. Kaplan LC, Matsuoka R, Gilbert EF, Opitz JM, Kurnit DM. Ectopia cordis and cleft sternum: evidence for mechanical teratogenesis following rupture of the chorion or yolk sac. Am J Med Genet 1985; 21: 187-202.