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Pentalogy of Cantrell accompanied by scoliosis and pes equinovarus deformity at 12 weeks gestation

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Summary

Cantrell's pentalogy (CP) is a rare syndrome characterized by defects in the lower sternum with ectopia cordis, anterior diaphragm defects, midline supraumbilical abdominal wall defects, defects in the diaphragmatic pericardium, and congenital heart disease. The authors report a 12-weeks gestation with multiple fetal anomalies suggesting the diagnosis of CP (a large thoraco-abdominal defect with herniating liver and bowel, heart deviated anteriorly with concomitant ventricular septal defect), and the 'S' shaped fetal spine due to increased lumbar lordosis and scoliosis with accompanying pes equinovarus deformity. Chorionic villus sampling was performed due to increased nuchal translucency (3.7 mm). The fetal karotype was found to be 47, XX,+21 (trisomy 21). In the literature, three scoliosis cases have been reported accompanying the CP along with multiple anomalies and one concomitant pes equinovarus deformity has been reported previously.

Key words: Pentalogy of Cantrell; Scoliosis; Pes equinovarus deformity; Prenatal diagnosis.

Introduction

Cantrell's pentalogy (CP) was first described in 1958 [1]. Cantrell Haller and Ravitch reviewed the literature to identify similar conditions and proposed five core features for this syndrome. CP is a rare combination of anomalies, comprising: a defect of the lower sternum, midline supraumbilical abdominal wall defect, deficiency of the anterior diaphragm, deficiency in the diaphragmatic portion of the pericardium with communication between the pericardial and peritoneal cavities, and congenital heart defect [1]. The full spectrum is estimated to have an incidence of 5.5 per one million live births [2]. There is male dominance with a male to female ratio of 2.7 : 1. With prenatal ultrasonography, the CP usually can be diagnosed in the first trimester of pregnancy [3]. Pentalogy is often incompletely present, with various combinations of two or three defects that are more common. The etiology and pathogenesis are still unknown and usually occurs as a sporadic event. Here, the authors presented an early prenatal diagnosis of incomplete CP with Trisomy 21 accompanying vertebral column anomalies and pes equinovarus deformity.

Case Report

A 23-year-old woman was referred at 12 weeks gestation with multiple fetal anomalies (a large thoraco-abdominal wall defect, heart deviated anteriorly). She had no significant obstetrical history. In the obstetric ultrasonography, single, living fetus with 60.5 mm crown-rump length and compatible with 12 weeks was detected. Ultrasonographic assessment identified a large exomphalos and inferior sternal defect, with the heart deviated anteriorly, which suggested the diagnosis of CP. The authors could not fully determine cardiac anomalies due to gestational age and deviated heart, but tricuspid regurgitation was detected and with transvaginal ultrasonography, a large ventricular septal defect was seen. The fetal spine could not be clearly evaluated due to increased lumbar lordosis and scoliosis. The vertebral column was 'S' shaped and was accompanied by pes equinovarus deformity. Nuchal translucency was also increased (3.7 mm).

Current findings and the prognosis of the disease were explained to the family. Chorionic villus sampling was performed. The genetic examination revealed a 47, XX, +21, (trisomy 21). The parents opted for termination. Pregnancy was terminated within 12 hours 600 microgram in total of vaginal misoprostol. In the macroscopic examination of female fetus, a wide median defect was seen, including abdominal wall and lower thorax. Intra-abdominal organs, liver and bowel, herniated by this defect, and anteriorly deviated heart were seen (Figure 1). The fetal spine was 'S' shaped due to increased lordosis and scoliosis. There was no concomitant neural tube defect however pes equinovarus deformity was seen in the right foot (Figure 2).

Discussion

CP is a rare syndrome characterized by defects in the lower sternum with ectopia cordis and various congenital heart diseases, anterior diaphragm defects, midline supraumbilical abdominal wall defects, and diaphragmatic pericardial defects.

In 1972, Toyoma, further classified the degree of expression of CP into three classes. Class I is a definite diagnosis with all five defects present. Class II is a probable diagno-

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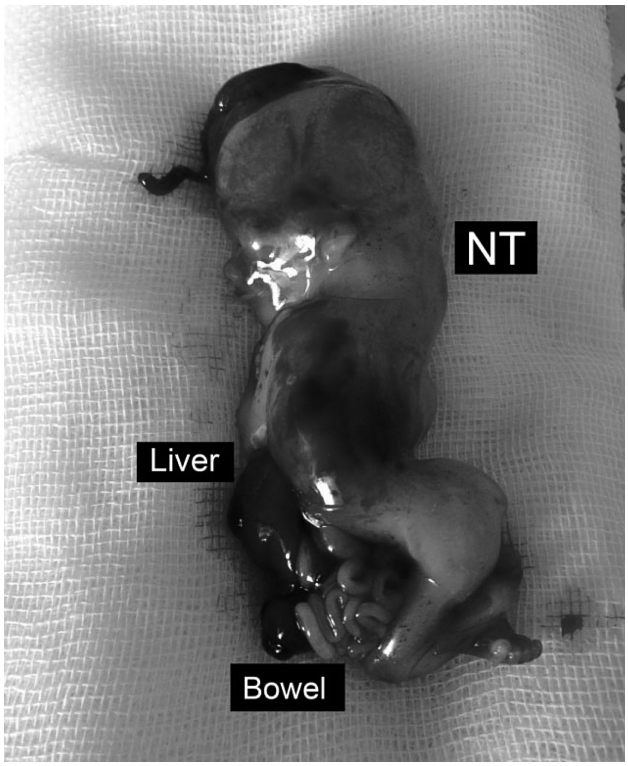

Figure 1. — Thoraco-abdominal wall defect and herniated organs with increased nuchal translucency.

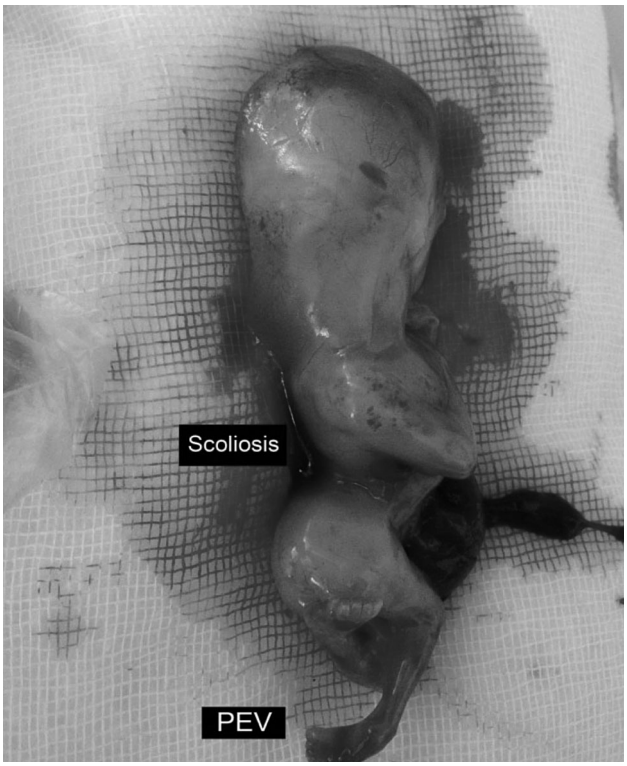

Figure 2. — Due to increased lordosis and scoliosis, 'S' shaped spine with pes equinovarus deformity.

sis with four of the five defects expressed, and Class III is considered an incomplete expression with varying combinations of defects present [4]. CP is often incompletely present, as seen in this case. The present case was represented with ectopia cordis, ventricular septal defect, supraumbilical abdominal wall defect, with herniated organs including liver and bowel. However lower sternal defect, anterior diaphragm defect, and diaphragmatic pericardial defect could not be evaluated due to gestational age. If pericardial effusion can be seen, associated anterior diaphragmatic hernia and diaphragmatic pericardial defects may be suspected, but the present fetus did not have pericardial effusion. The etiology and pathogenesis of the CP are still unknown. The hypothesis underlying this condition probably originates between the 14 and 18 days of embryonic life, with an alteration in the migration of the primordial mesodermic structures of the medial line [5]. This results with failure of ventral wall closure and incomplete external primordial bands fusion. Incomplete closure and fusion would account for defects in the chest and abdominal wall, aplasia of the anterior diaphragm, and the associated pericardium. Organs may eviscerate through the resulting sternal and abdominal wall defects. Ectopia cordis is characterized by complete or partial displacement of the heart outside the body. Intracardiac anomalies are described in CP including the most common ventricular septal defect, but also tetralogy of Fallot, atrial septal defects, and ventricular diverticulum [1]. Several cases of the CP reported in the literature had central nervous system and craniofacial malformations, such as cleft lip and palate, craniorachischisis, encephalocele, and hydrocephalus [6-8]. Various other associated anomalies have been reported; limb defects such as clubfoot, absence of tibia or radius, hypodactyly and abdominal organ defects such as gallbladder agenesis and polysplenia [9-11]. This syndrome has also been reported occasionally in association with sirenomelia. In the present case the fetus also had increased lordosis and severe scoliosis concomitant with pes equinovarus deformity. In the literature, three scoliosis cases have been reported accompanying CP, along with multiple anomalies and one concomitant pes equinovarus deformity has been reported [12, 13].

Most cases are believed to occur sporadically and no recurrences have been reported. Although sporadic in most of the described infants, X-linked recessive inheritance was suggested for some families, and genes located on the X-chromosome (Xq25-q26.1) may be involved in some of the reported cases [5]. There are much suspicious conditions as chromosome imbalances, an early amniotic sac rupture, and amniotic bands [14, 15].

Prenatal diagnosis is important because the prognosis is poor and the disease is lethal. With prenatal high resolution ultrasound and fetal magnetic resonance imaging, CP can

usually be diagnosed in the first trimester of pregnancy as in the present case [3]. However, diagnosis can be especially established during the second trimester in 60% of cases with incomplete defects [16].

Chromosomal abnormalities especially should be kept in mind and with increased nuchal translucency measurement prenatal diagnostic methods should be performed. In the present case, chromosome examination result was found as 47, XX,+21 but most frequently defined chromosomal anomaly associated with pentalogy is Trisomy 18 which can be found in 5-10% of cases [17].

Although survival of the CP depends on the size of the abdominal wall defect, extent of the cardiac defect, and presence of associated anomalies, because of the poor prognosis, termination of pregnancy should be offered in cases diagnosed at early weeks.

In conclusion, presence of fetal abdominal wall defects and increased nuchal translucency in ultrasonography should be carefully investigated for the diagnosis of CP and chromosomal anomalies. As in the present case, CP can be accompanied by chromosomal abnormality. Therefore early diagnosis is important for making appropriate counseling to the family and aid in decision-making regarding pregnancy.

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