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Case Report

Prenatal diagnosis of complete Cantrell's pentalogy in an IVF/ICSI co-twin

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Abstract

The pentalogy of Cantrell (POC) is a rare syndrome with an incidence of 5.5 per 1 million live births with an unknown etiology. POC includes deficiency of the anterior diaphragm, defect in the supraumbilical abdominal wall in the midline and in the diaphragmatic pericardium, and also of the lower sternum and intracardiac abnormality. It has been suggested that the risk of birth defects are slightly increased in infants conceived by assisted reproductive technologies. Here, we reported the prenatal diagnosis of complete POC in an IVF/ICSI co-twin in the first trimester using two-dimensional ultrasound.

Key words:

Fetal, pentalogy of Cantrell, prenatal, twin, ultrasound

Introduction

The pentalogy of Cantrell (POC) is a rare syndrome with an incidence of 5.5 per 1 million live births with an unknown etiology. There is a male dominance with a male to female ratio of 2,7:1. POC includes deficiency of the anterior diaphragm, defect in the supraumblical abdominal wall in the midline and in the diaphragmatic pericardium, and also of the lower sternum and intracardiac abnormality. There are cases with the complete syndrome and incomplete variants [1]. Prognosis of the disease depends on the severity of the abnormality, and is usually poor. It has been suggested that the risk of birth defects are slightly increased in infants conceived by assisted reproductive technologies (ART) [2-5]. Here, we reported the prenatal diagnosis of complete POC in an IVF/ICSI co-twin in the first trimester using two-dimensional ultrasound.

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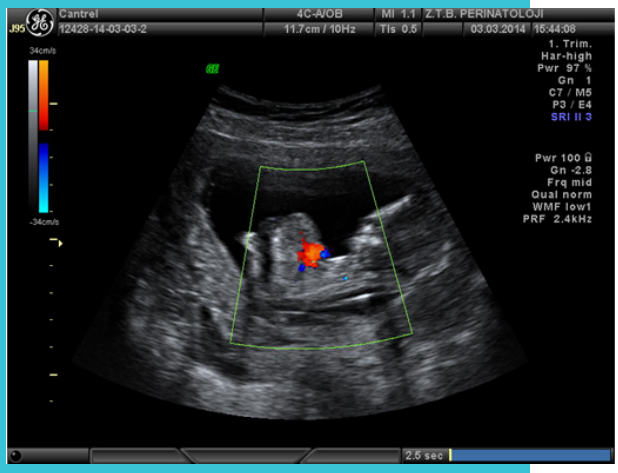
Case presentation

A 31-year-old primigravid woman with a diamniotic and dichorionic twin pregnancy, was 14 weeks admitted to the outpatient clinic for a routine first trimester control. In her past history, she described male infertility, an in vitro conception following intracytoplasmic sperm injection and transfer of three embryos that ended up with twins, one of which was lost at 9 weeks of gestation and also a consanguineous marriage between first cousins. She reported taking no medications except folic acid. No history of any genetic or structural anomalies was noted in herself, her husband, and in the family. The two dimensional ultrasound at 14 weeks revealed a live fetus with an increased nuchal translucency (NT) of 4,6 mm, a large omphalocele, ectopia cordis, sacral myelomeningocele and a marked spinal curvature (Figure 1). After discussion with the family, without considering any further genetic testing, they decided to termination of the

pregnancy that was achieved by induction with misoprostol at 15 weeks of gestation. Post abortion examination and histopathologic evaluation at autopsy, in accord with the prenatal sonographic findings, revealed a large anterior thoraco-abdominal wall defect, an absence of anterior diaphragm and pericardium, an omphalocele containing an edematous liver and spleen, also loops of small intestine and colon, an ectopic heart with ventricular septal defect, a sacral meningocele, and a normal male phenotype with a karyotype of 46, XY (Figure 2). A plain radiography demonstrated severe thoracolumbar lordoscoliosis, 27 total number of vertebrae, absence of sternum and coccyx, absence of fibula on the right, and distortion of the femoral head on both sides (Figure 3).

Figure 1.

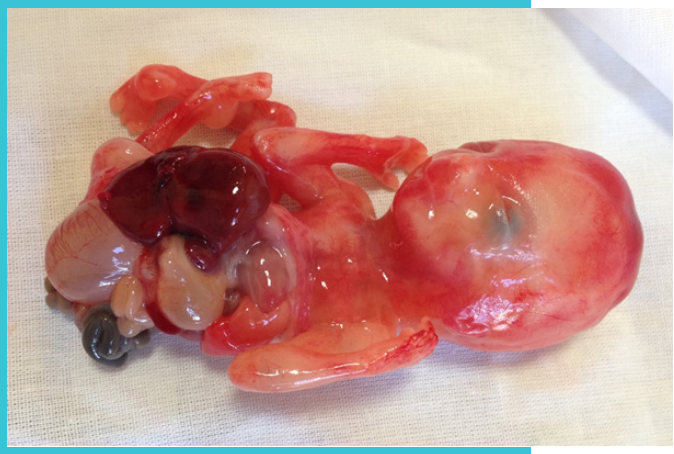


Two-dimensional colour doppler sonogram demonstrates a large omphalocele with ectopia cordis

Discussion

Although the etiology is unknown, a developmental failure of the mesoderm prior to differentiation into the somatic and splanchnic layers, between 14 and 18 days after conception has been hypothesized to result in the defects involved in the sternum, abdominal wall, pericardium, diaphragm and the myocardium, as seen in POC. Although most cases are sporadic, an association with trisomy 18, 13, Turner syndrome and X-linked inheritance has been described. The differential diagnosis in POC includes isolated ectopia cordis, body-stalk anomaly, amniotic band syndrome and simple omphalocele [6-8]. The association between the birth defects and ART has been attributed to the IVF/ICSI procedures, perhaps due to the bypassed natural selection of the gametes, the early embryonic development taking place at the laboratory and later in the uterus in a hyperstimulated hormonal environment. The parents' underlining infertility also is associated with the increased birth defects [2, 4]. An increased risk of abdominal wall defects including omphalocele has been reported with and without IVF/ICSI, especially in multiple pregnancies, in subfertile patient groups [5, 9, 10].

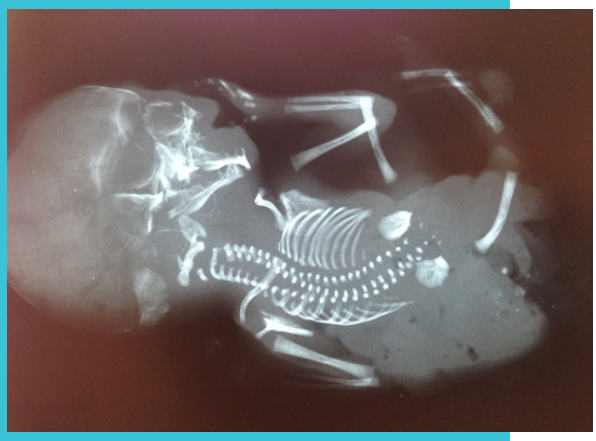
Figure 2.



The features of the pentalogy of Cantrell in the abnormal co-twin

When congenital malformations occur in a twin pregnancy, management decisions can be difficult especially in monochorionic ones. The main fetal intervention available for twin gestations is selective termination of anomalous co-twin by several techniques including cardiac puncture, intracardiac injection of potassium chloride or cord occlusion. Determining chorionicity is vital prior to consider selective termination [9,10]. The association of POC with increased nuchal translucency, as in our case, may be caused by venous congestion due to the cardiac defects and displacement, also to mediastinal compression by the diaphragmatic hernia, or omphalocele [11]. Likewise, skeletal manifestations of our case were not unique. Some cases may have associated midline defects also such as cleft lip or palate, exenceph

Figure 2.



The plain radiography demonstrating the thoracolumbar lordoscoliosis and multiple skeletal defects

aly, sirenomelia or other skeletal abnormalities [1,12, 13]. The combination of omphalocele and ectopia cordis is the major hallmark of POC that makes the definite diagnosis of the syndrome feasible at an early stage of gestation, like at the end of the first trimester by ultrasound. In complete expression of the syndrome like in the case we presented here, the prognosis is very poor and the disease is lethal, therefore early prenatal diagnosis and termination of pregnancy is feasible and extremely important.

Acknowledgements

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Conflict of Interest

None

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