Case Report

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First trimester diagnosis of body stalk anomaly complicated by ectopia cordis

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Abstract

Body stalk anomaly is a rare abnormality characterized by an abdominal wall defect with evisceration of abdominal organs, severe kyphoscoliosis, and a very short or absent umbilical cord. Ectopia cordis (EC) is a rare, lethal anomaly characterized by complete or partial malpositioning of the heart outside of the thorax. A 28-year-old healthy primigravida was referred to our department to undergo a nuchal translucency thickness scan at 12 weeks' gestation. The scan revealed typical features of body stalk anomaly and EC. Given the lethal condition of the fetus, the patient opted for termination of the pregnancy. Body stalk anomalies, especially those complicated by EC, are universally lethal for the affected fetus. Selective termination should be recommended to avoid possible complications that can arise during pregnancy. Additionally, the future parents should be informed that because the condition is not associated with chromosomal abnormalities, there is no increased risk of recurrence.

Keywords

Body stalk anomaly, ectopia cordis, termination of pregnancy, complex body wall anomalies, transvaginal ultrasound, case report

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Introduction

Body stalk anomaly (BSA) and limb–body wall complex (LBWC) are examples of complex body wall anomalies. Typically, BSA is considered when clinical examination reveals a large abdominal wall defect with protrusion of viscera, severe kyphoscoliosis, and a very short or absent umbilical cord (UC), often continuous with the placenta. The pivotal features of LBWC are body wall and structural limb anomalies with or without craniofacial abnormalities.[1] The reported frequency of BSA is 1 per 7500 first-trimester pregnancies.[2] Various theories have been proposed to explain the possible pathogenesis of this anomaly, such as abnormal early embryonic folding,[3] early amnion rupture,[4] teratogenic exposure in early pregnancy,[5] and vascular disruption of the early embryo.[6] However, the actual etiology remains unknown. Most of the described cases had a normal karyotype; only two cases had chromosomal abnormalities associated with uniparental disomy of chromosome 16 and a chromosomal trisomy, respectively.[7] With the profound implementation of sonographic examination during pregnancy and our increasing understanding of this anomaly, BSA can typically be diagnosed in the first trimester. The differential diagnoses include an isolated omphalocele or gastroschisis, short UC syndrome, and other polymalformative syndromes. In light of the multiple abnormalities associated with BSA, selective termination is performed in most prenatally diagnosed cases; however, occasional cases of long-term survival have been reported. Ectopia cordis (EC) is defined as complete or partial displacement of the heart outside the thoracic cavity. Ultrasound diagnosis of this anomaly is confirmed by observing a pulsating heart that is partly or completely outside the thoracic cage. Diagnosis of EC may be difficult because only part of the heart is extrathoracic, and visualization is hindered by protruding abdominal contents. We herein report a case of BSA complicated by EC diagnosed at 12 weeks of gestation.

Case report

A 28-year-old healthy primigravida was referred to our department to undergo a nuchal translucency thickness scan at 12 weeks' gestation. Her family history was unremarkable, and she had no history of drug abuse or infectious disease. A detailed transabdominal ultrasound scan revealed a live fetus with a crown–rump length of 4.7 cm, consistent with a gestational age of 12 weeks. The nuchal translucency thickness was 0.15 cm.

A large anterior abdominal wall defect and deficiency of the left leg were noted. Amniotic membrane-like echo was visible in the uterus. The patient was further evaluated using three-dimensional transvaginal ultrasound, which revealed a defect in the anterior abdominal wall with exposure of the bowel and liver. A short UC was visualized. Other anomalies included scoliosis, the pulsating heart outside the thorax, and right talipes equinovarus. The amniotic fluid volume was normal (Figure 1). Based on these examination findings, a diagnosis of BSA with EC was considered.

Given the lethal condition of the fetus, the patient opted for termination of the pregnancy, and an uncomplicated procedure was performed. Postmortem examination of the fetus demonstrated eviscerated abdominal organs adherent to bands of amniotic tissue, protrusion of the heart out of the thoracic cavity, a short UC of 15 mm, and amelia, which were consistent with the ultrasound findings (Figure 2). Fetal karyotyping was recommended, but the parents declined.

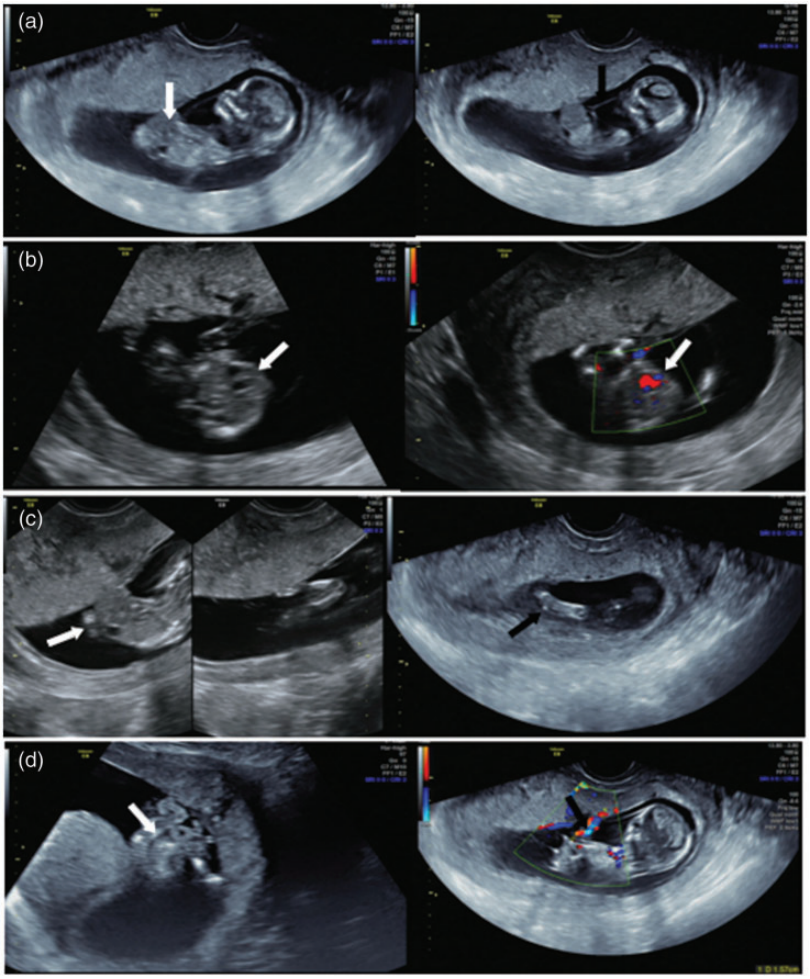

Figure 1. Sonogram of the fetus at 12 weeks' gestation. (a) Defect of the anterior abdominal wall (white arrow) and amniotic membrane-like echo in the uterus (black arrow). (b) Pulsation of the heart outside the thorax (white arrow). (c) Amelia (white arrow) and right talipes equinovarus (black arrow). (d) Severe kyphoscoliosis of the lower spine (white arrow) and a very short umbilical cord (black arrow).

Discussion

BSA was first reported in 1906 as a pathologic entity in a neonate with an abdominal wall defect, evisceration of abdominal organs, and an absent UC.[8] Because of the diverse phenotypes among reported cases, terms such as amniotic band syndrome and LBWC have also been used to describe the condition. Van Allen et al.[6] proposed that a diagnosis of BSA should meet at least two of the following three criteria: exencephaly/encephalocele with facial clefts, thoracoschisis and abdominoschisis (midline defect), and a limb defect (e.g., clubfoot, polydactyly, oligodactyly, syndactyly, brachydactyly, or amelia). Martín-Alguacil and Avedillo[9] recently proposed the following classification of BSA based on anatomical and embryological criteria

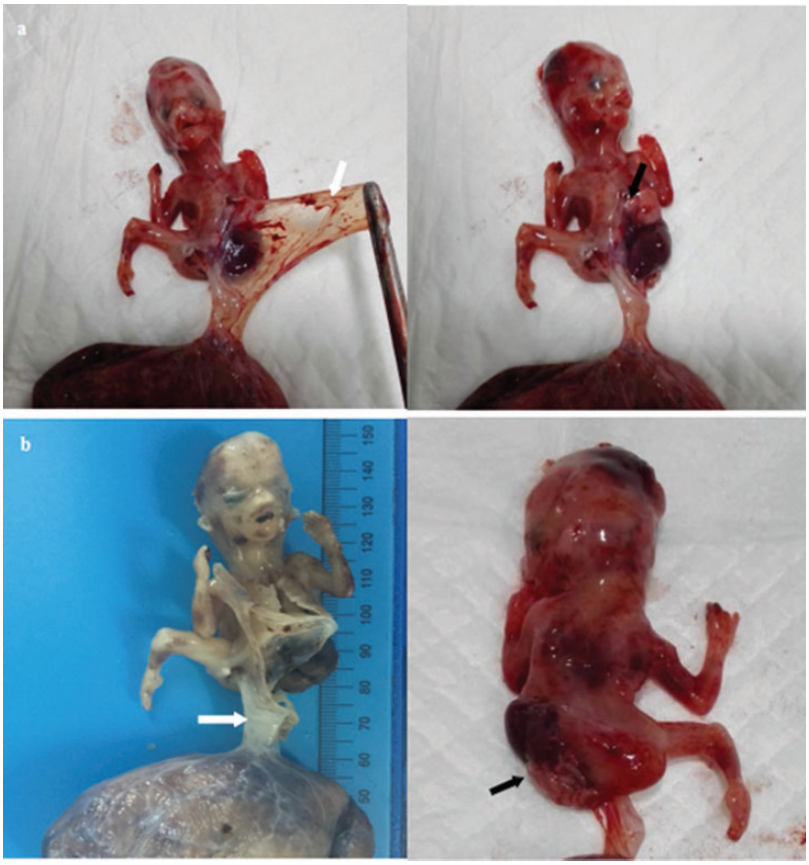

Figure 2. Postmortem examination of the fetus. (a) Eviscerated abdominal organs adherent to bands of amniotic tissue (white arrow) and protrusion of the heart out of the thoracic cavity (black arrow). (b) Short umbilical cord of 15 mm (white arrow) and amelia.

in a pig model: (a) BSA Type I: fetus with spinal and UC defects, thoracoabdominoschisis, anal atresia and/or other internal organ structural defects, and structural limb defects; (b) BSA Type II: fetus with spinal and UC defects, thoracoabdominoschisis, anal atresia and/or other internal organ structural defects, and nonstructural limb defects; (c) BSA Type III: fetus with spinal and UC defects, abdominoschisis, anal atresia and/or other internal organ structural defects, and structural limb defects; and (d) BSA Type IV: fetus with spinal and UC defects, abdominoschisis, anal atresia and/or other internal organ structural defects, and nonstructural limb defects. A prenatal diagnosis of BSA can be made as soon as 9 weeks,[10] and all cases can be identified during the first trimester in centers with experienced sonographic clinicians.

EC is a rare, lethal anomaly characterized by complete or partial malpositioning of the heart outside of the thorax, and its prevalence is 5 to 8 per million deliveries. EC is classified into four subgroups according to the position of the heart: 3% of cases are cervical, in which the heart is displaced superiorly into the neck; 60% of cases are thoracic, in which the heart protrudes anteriorly through a sternal defect; 7% of cases

are thoracoabdominal, in which the heart is displaced outside the chest through a defect in the lower sternum in association with diaphragmatic and ventral abdominal wall defects; and 30% of cases are abdominal, in which the heart is displaced into the abdomen through a defect in the lower sternum and diaphragm.[11] Although isolated EC has been reported, the anomaly frequently occurs in conjunction with pentalogy of Cantrell, a combination of anomalies comprising EC, sternal clefts, ventral diaphragmatic hernias, abdominal wall defects, and cardiac anomalies.

Because of the rarity of BSA and EC, the existing literature consists predominantly of case reports and hospital-based case series identified retrospectively over many years; additionally, descriptions of the relationship between the two conditions are inconsistent. Bugge[12] reported 16 cases of EC in a 20-year epidemiological study of BSA from 1970 to 1989 in Denmark. Of these 16 cases, only 3 exhibited thoracoabdominoschisis, whereas urogenital anomalies occurred in 10 cases.[12] Additionally, Smrcek et al.[13] reviewed 11 cases of BSA, among which 4 (36%) cases were associated with EC. In contrast, however, Sepulveda et al.[14] reported that two (50%) of four cases of BSA were associated with EC. This discrepancy can be explained by a fetus with BSA and EC being not likely to survive the first trimester of pregnancy, and most such cases result in spontaneous miscarriage without detection. Moreover, the association between the two defects might be underestimated because of the low detection rates of thoracoabdominal EC. Therefore, it is reasonable to postulate that the incidence of EC may not be rare in cases of BSA.

The present case demonstrates the typical features of BSA and thoracic EC in a fetus. The case was classified as Type I according to the BSA classification system recently described by Martín-Alguacil and Avedillo,[9] and it corresponded to LBWC Type I characterized by thoracoabdominoschisis and structural limb anomalies. Our findings are in agreement with the hypothesis that early amniotic rupture before obliteration of the coelomic cavity causes BSA. An alternative hypothesis suggested by Streeter[3] can also account for our findings. It is theorized that during the very early stages of human development (4th to 5th weeks), the folding of the embryo progresses abnormally; specifically, all of the axes (cephalic, caudal, and lateral) fold, and the flat trilaminar embryo is thus transformed into a cylindrical fetus resulting in coelomic cavity obliteration failure and dysgenesis of the UC. A third hypothesis for the pathogenesis of BSA is early comprehensive compromise of embryonic blood flow. In animal studies, the rat peripheral vasculature underwent disturbances when the early amniotic membrane was artificially ruptured.[15]

BSA, especially when complicated by EC, is universally lethal for the affected fetus. As soon as a prenatal diagnosis is made, selective termination is the best option to avoid possible complications that can arise during pregnancy. Additionally, the future parents should be informed that because the condition is not associated with chromosomal abnormalities, there is no increased risk of recurrence.

Declaration of conflicting interest

The authors declare that there is no conflict of interest.

Ethics

This report was approved by the ethics committee of The First Hospital of Jilin University. The patient provided written consent for publication of this case report in accordance with the principles of the Declaration of Helsinki.

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References

1. Martin-Alguacil N. Anatomy-based diagnostic criteria for complex body wall anomalies (CBWA). *Mol Genet Genomic Med* 2020; 8: e1465. DOI: 10.1002/mgg3.1465.
2. Daskalakis G, Sebire NJ, Jurkovic D, et al. Body stalk anomaly at 10–14 weeks of gestation. *Ultrasound Obstet Gynecol* 1997; 10: 416–418.
3. Streeter GL. Focal deficiencies in fetal tissues and their relation to intrauterine amputations. *Contrib Embryol* 1930; 22: 1–44.
4. Torpin R. Amniochorionic mesoblastic fibrous strings and amnionic bands: associated constricting fetal malformations or fetal death. *Am J Obstet Gynecol* 1965; 91: 65–75.
5. Herva R and Karkinen-Jääskeläinen M. Amniotic adhesion malformation syndrome: fetal and placental pathology. *Teratology* 1984; 29: 11–19.
6. Van Allen MI, Curry C and Gallagher L. Limb body wall complex: I. Pathogenesis. *Am J Med Genet* 1987; 28: 529–548. DOI: 10.1002/ajmg.1320280302.
7. Chan Y, Silverman N, Jackson L, et al. Maternal uniparental disomy of chromosome 16 and body stalk anomaly. *Am J Med Genet* 2000; 94: 284–286.
8. Iba T, Harada T, Iba Y, et al. Concordant body stalk anomalies in dichorionic twins. *J Ultrasound Med* 2016; 35: 2736–2739.
9. Martín-Alguacil N and Avedillo L. Body stalk anomalies in pig—definition and classification. *Mol Genet Genomic Med* 2020; 8: e1227.
10. Quijano FE, Rey MM, Echeverry M, et al. Body stalk anomaly in a 9-week pregnancy. *Case Rep Obstet Gynecol* 2014; 2014: 357285.
11. Turkyilmaz G, Avci S, Sivrikoz T, et al. Prenatal diagnosis and management of ectopia cordis: varied presentation spectrum. *Fetal Pediatr Pathol* 2019; 38: 127–137. DOI: 10.1080/15513815.2018.1556367.
12. Bugge M. Body stalk anomaly in Denmark during 20 years (1970-1989). *Am J Med Genet A* 2012; 158A: 1702–1708. DOI: 10.1002/ajmg.a.35394.
13. Smrcek JM, Germer U, Krokowski M, et al. Prenatal ultrasound diagnosis and management of body stalk anomaly: analysis of nine singleton and two multiple pregnancies. *Ultrasound Obstet Gynecol* 2003; 21: 322–328. DOI: 10.1002/uog.84.
14. Sepulveda W, Wong AE and Fauchon DE. Fetal spinal anomalies in a first-trimester sonographic screening program for aneuploidy. *Prenat Diagn* 2011; 31: 107–114. DOI: 10.1002/pd.2608.
15. Daskalakis GJ and Nicolaides KH. Monozygotic twins discordant for body stalk anomaly. *Ultrasound Obstet Gynecol* 2002; 20: 79–81.