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Body stalk anomaly: antenatal sonographic diagnosis of this rare entity with review of literature

Amandeep Singh, Jasmeet Singh, Kamlesh Gupta

SGRD Institute of Medical Sciences and Research, Amritsar, India
Department of Radiodiagnosis and Imaging, Sri Guru Ramdas Institute Of Medical Sciences And Research, Vallah, Amritsar

Correspondence: Dr Amandeep Singh, Department of Radiodiagnosis and Imaging, Sgrdimsar, Vallah, Amritsar, 469, East Mohan Nagar, Sultanwind Road, Amritsar-Punjab 143001, India, tel.: +91 9872454954, e-mail: dr.amancs@gmail.com

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Abstract

Body stalk anomaly is a rare and severe malformation syndrome in which the exact pathophysiology and trigger factors are still unknown. Possible causes of body stalk anomaly include early amnion rupture with direct mechanical pressure and amniotic bands, vascular disruption of the early embryo, or an abnormality in the germinal disk. We present a case of body stalk anomaly diagnosed during antenatal sonographic evaluation at the first visit with the review of literature regarding this phenomenon. Sonographic features of the fetus included a severe midline defect of the fetal abdominal wall with a large extra-abdominal mass containing bowel and liver inside. Body stalk anomaly is accepted as a fatal anomaly, so it is important to differentiate it from other anterior wall defects for evaluating the management options.

Introduction

Anterior abdominal wall defects are roughly classified into three types: gastroschisis, omphalocele and body stalk-like anomalies[1]. Body stalk anomaly is the rarest, most severe and invariably lethal abdominal wall defect. It is a severe defect in which the abdominal wall does not develop and thus the peritoneal cavity is open to the extraembryonic coelom and the fetus is attached to the placenta[1]. The presence of the liver and intestine in the extraembryonic coelom differentiates body stalk anomalies from other subtypes. Body stalk anomalies are generally not associated with chromosomal anomalies[1]. Likewise, this anomaly might also occur in conjunction with neural tube defects, genitourinary malformations, abnormalities of the chest wall, intestinal atresia, and craniofacial defects, among others[2,3]. The variety of phenotypes in the reported cases worldwide has led to the creation of a confusing array of terms for this condition including the amniotic band syndrome, short umbilical cord syndrome, and limb-body wall complex[4]. This rare malformation syndrome has a reported prevalence of 0.12 cases per 10,000 births (including both live and still births)[5,6]. However, in a recent multicenter study of Daskalakis *et al*[1], in which 106,727 fetuses between 10 and 14 weeks of gestation were analyzed, an incidence of 1/7,500 pregnancies was found. This discrepancy in the incidence rates suggests that this type of malformation might be responsible for a significant number of spontaneous abortions during the first trimester of pregnancy, and thus the real incidence for this anomaly might be underestimated.

Case presentation

A 27-year-old healthy primigravida of Indian origin presented to Obstetrical OPD for routine antenatal checkup. The gestational history was uneventful with no other relevant past medical or surgical history. Her menstrual cycle was regular. Her gestational age according to the last menstrual period was 15 weeks. Her initial prenatal tests were within the normal limits. Grey scale ultrasound revealed the mean gestational age to be 15 weeks 3 days. The fetus had a large abdominal wall defect and ventriculomegaly with thinned out cerebral parenchyma. The patient was further evaluated using three-dimensional ultrasound and color doppler. There was gross dilatation of bilateral lat-

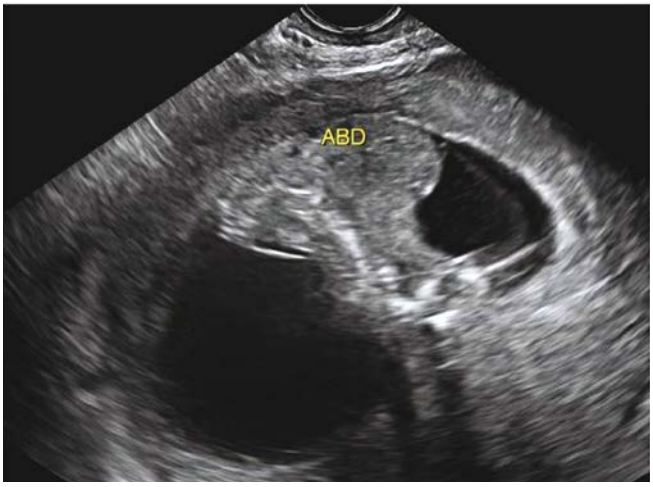

Fig. 1. *Ultrasound image shows herniated abdominal (ABD) contents attached to the placenta*

eral ventricles and cerebral parenchyma was thinned out and compressed. The cerebellum appeared compressed against the occiput. Also seen was a large abdominal wall defect with herniation of viscera, i.e. the liver and bowel loops through the defect suggesting a large omphalocele (Fig. 1, Fig. 2). However, other abdominal organs including the stomach, spleen and urinary bladder were within the body. The herniated viscera seemed to be attached to the placenta (Fig. 1, Fig. 2). The heart was in the normal location within the thoracic cavity. Kyphoscoliosis of the visualized spine was seen. The amniotic fluid was normal. Three dimensional USG revealed attachment of herniated viscera to the placenta and confirmed the diagnosis of body stalk complex. Alpha-fetoprotein levels were raised. Because the malformation is incompatible with life, the patient opted for termination of pregnancy.

Examination of the fetus revealed a large omphalocele with herniation of the small intestine and liver. A very short umbilical cord was observed which was attached to the herniated organs (Fig. 3). The herniated organs were covered by the amniotic membrane and attached to the placenta.

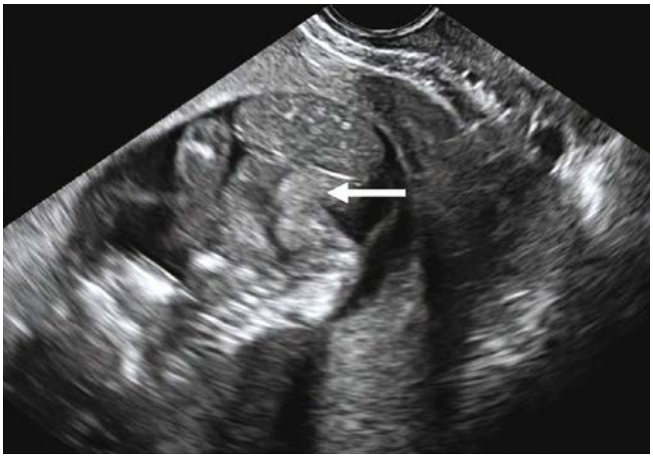

Fig. 2. *Ultrasound image shows abdominal wall defect with herniation of the abdominal viscera (arrow)*

Discussion

Body stalk anomaly is a term used to describe a pattern of severe defects that in most of the reported cases proves to be incompatible with life. This condition should be suspected when a large abdominal defect as well as abnormalities in the axial skeleton such as kyphosis or scoliosis are observed, and a short or absent umbilical cord is found. Body stalk defects can be detected at the end of the first trimester of pregnancy by ultrasound. It is also important to consider other pathologies that affect the abdominal wall such as omphalocele, gastroschisis, bladder exstrophy, cloacal exstrophy, Cantrell pentalogy, and the OEIS complex (omphalocele, exstrophy of cloaca, imperforate anus, and spinal defects)[6].

Possible causes of body stalk anomaly include early amnion rupture with direct mechanical pressure and amniotic bands, vascular disruption of the early embryo, or an abnormality in the germinal disk[7]. Defects in genes related to embryogenesis may play a role[8].

The germinal disk abnormality is thought to represent complete failure of body folding along all three axes (ce-

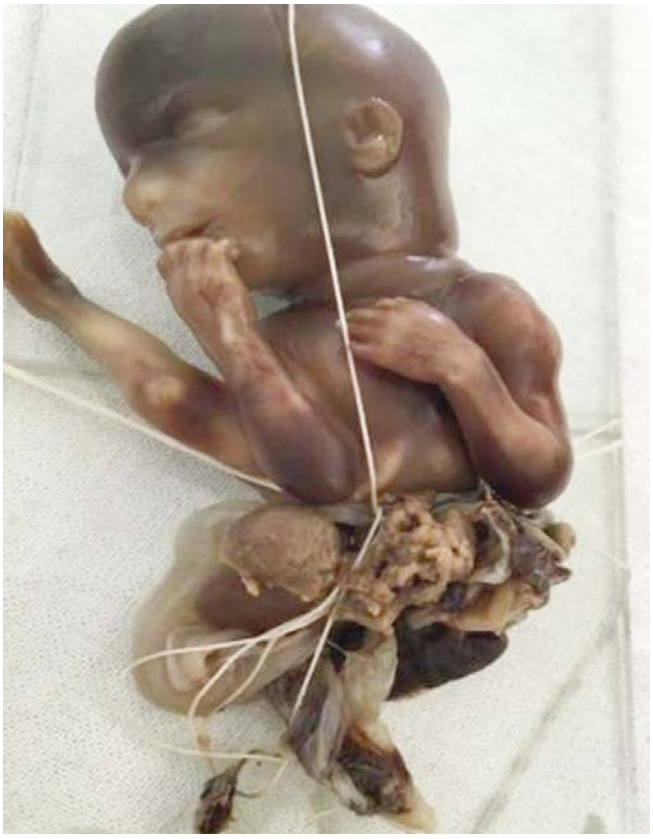

Fig. 3. *Fetus with large omphalocele with herniation of the small intestine and liver. A short umbilical cord was observed which was attached to the omphalocele*

phalic, caudal, and lateral)[9]. Normal body folding results in separation of the intraembryonic coelom (future peritoneal cavity) from the extraembryonic coelom, formation of the body stalk, and development of the umbilical cord[10,11]. Aberrant cephalic folding leads to a defect in the thoracic wall and epigastrium, which allows development of ectopia cordis. Aberrant lateral folding results in herniation of the midabdominal contents into a large wide-based amnioperitoneal sac, which inserts peripherally onto the placental chorionic plate in lieu of an umbilical cord or with a very short umbilical cord[10,11].

Due to the extrusion of the intra-abdominal contents, the spine and thoracic cavity do not develop symmetrically, which results in severe scoliosis and abnormalities of the axial skeleton. Malrotation of the spine and incomplete closure of the pelvis can lead to malrotated limbs and/or club feet[10,11].

Van Allen et al. set forth the diagnostic criteria for BSA in 1987. Two of the three following anomalies must be presented to establish a positive diagnosis[7,12]:
- Exencephaly/encephalocele with facial clefts
- Thoraco- and abdominoschisis (midline defect)
- Limb defect (i.e. club foot, polydactyly, oligodactyly, syndactyly, brachydactyly, amelia).

Two main phenotypes have been described in the literature ([13,14], each being the consequence of different pathogenic mechanisms[12]:
- The placental-cranial type which involves craniofacial defects (encephalocele/exencephaly associated with facial clefts) and amniotic bands between the cranial defects and placenta – the pathogenic mechanism proposed is early vascular disruption.
- The placental-abdominal type in which no craniofacial defects are present, but which involves urogenital anomalies, anal atresia, lumbosacral meningocele, short cord, persistence of extraembryonic coelom and intact amnion – it seems to be due to intrinsic abnormal embryonic development.

In our case, the ultrasonographic findings were consistent with those reported in the literature.

Nonetheless, it is essential to make an early diagnosis in order to provide the future parents with the necessary information and counseling regarding the prognosis of this type of anomaly. It is also important to remember that there are no specific therapeutic interventions for the fetus that usually dies shortly after delivery.

Conflict of interest

The authors do not report any financial or personal connections with other persons or organizations, which might negatively affect the content of this publication and/or claim authorship rights to this publication.

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