ISSN 2598-3180 (Online)
JOURNAL OF MIDWIFERY
Research and Practice
Accredited by SINTA 4th

Article

Body Stalk Anomaly: Antenatal Sonographic Diagnosis

Reyhan Julio Azwan [1], Bobby Indra Utama[2], Hudilla Rifa Karmia [2] Muhammad Iqbal[2], Yusrawati[2],Joserizal serudji [2]

[1]Departement of Anatomy, Faculty of Medicine, Andalas University, Indonesia
2Departement of obstetrics and Gynaecology, Faculty of Medicine, Andalas University, Indonesia

SUBMISSION TRACK

Recieved: Mei 07 2022
Final Revision: June 14 2022
Available Online: June 29 2022

KEYWORDS

body stalk anomaly, abdominal defect, ultrasound, limb body wall complex

CORRESPONDENCE

Phone: 0811668272
E-mail: yusrawati_65@yahoo.co.id

A B S T R A C T

The abstract should summarize the content of the paper. Try to keep the abstract below 200 words. Do not make references nor display equations in the abstract. The journal will be printed from the same-sized copy prepared by you. Your manuscript should be printed on A4 paper (21.0 cm x 29.7 cm). It is imperative that the margins and style described below be adhered to carefully. This will enable us to keep uniformity in the final printed copies of the Journal. Please keep in mind that the manuscript you prepare will be photographed and printed as it is received. Readability of copy is of paramount importance.

I. INTRODUCTION

Anterior abdominal wall defects are roughly classified into three types: gastroschisis, omphalocele and body stalk-like anomalies (Daskalakis et al, 2003) . 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(Daskalakis et al, 2003). 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 (Tsirka et al, 2007). 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 (Tsirka et al, 2007). 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 (Bianchi et al, 2000). This rare malformation syndrome has a reported prevalence of 0.12 cases per 10,000 births (including both live and still births). However, in a recent multicenter study of Daskalakis *et al* ,2003 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 REPORT:

A 29 years old healthy primigravida presented to OBGYN for routine antenatal check up. The gestational history was un eventful with no other relevant past medical or surgical history. Her menstrual cycle was regular. Her gestational age according to the last menstrual period was 35 weeks. Her initial prenatal tests were within the normal limits. Grey scale ultrasound revealed the mean gestational age to be 35 weeks 3 days. The fetus had a large abdominal wall defect. The patient was further evaluated using three-dimensional ultrasound and color doppler. The large abdominal wall defect with herniation of viscera, i.e. the liver and bowel loops through the defect suggesting a large omphalocele **(Fig. 1).** 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. 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. Because the malformation is incompatible with life, the patient opted for termination of pregnancy.

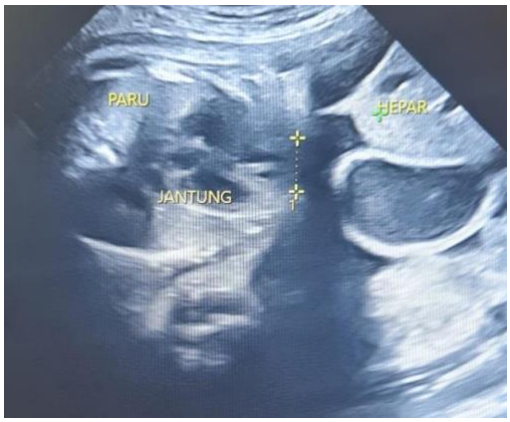

Figure.1 Ultrasound image shows herniated abdominal (ABD) contents attached to the placenta

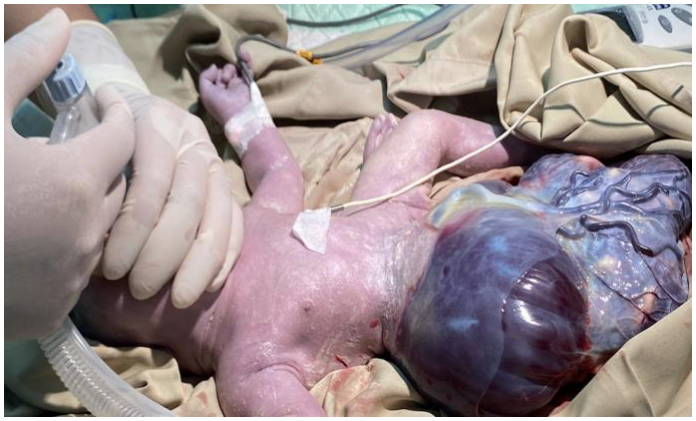

Figure.2 The image shows abdominal wall defect with herniation of the abdominal viscera (arrow)

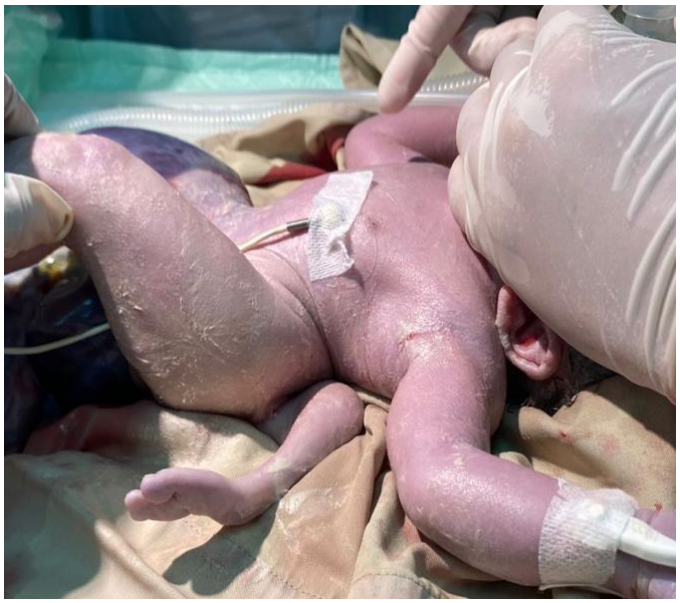

Figure.3 Examination of the fetus revealed a large omphalocele with herniation of the small intestine and liver. A very short umbilical cord was observed which was attached to the herniated organs **(Fig.2, Fig. 3).** The herniated organs were covered by the amniotic membrane and attached to the placenta.

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. (Bianchi et al, 2000).

The findings of this study highlight the pathognomonic features of BSA diagnosed during second trimesters of pregnancy. The main sonographic findings in association with BSA are large abdominal wall defect with abdominal organs attached to the placenta, absent umbilical cord and severe kyphoscoliosis; these findings have been previously described (Tsirka et al, 2007, Bianchi et al, 2000, Russo et al, 1993).

An additional finding from this case in relation to BSA is that the amniotic membrane was intact on its entire surface apart from the level of abdominal wall defect. The abdominal organs were herniated into the coelomic cavity and the rest of the fetus was located in the amniotic sac within a normal amount of amniotic fluid. The abdominal placental attachment was easily recognized in the first trimester as compared to later in pregnancy. Some authors have reported that in cases of BSA, the umbilical cord is very short (Daskalakis et al, 2003,Tsirka et al, 2007).

Differential diagnosis of abdominal wall defects in cludes exomphalos, gastroschisis, cloaca extrophy and OEIS complex, pentalogy of Cantrell, abdominoschisis due to amniotic bands and BSA. Figure 2 illustrates these conditions. Abdominal organs attached to the placenta, severe kyphoscoliosis and absence of a free floating umbilical cord are diagnostic for BSA. In exomphalos, the herniated viscera appear in the base of the umbilical cord and a freefloating cord is visible in the amniotic cavity. In gastroschisis, pentalogy of Cantrell and in cloaca extrophy, the eviscerated organs are within the amniotic cavity and the umbilical cord is freefloating. In abdominoschisis due to amniotic bands, the amniotic membrane continuity is lost, but the umbilical cord is free floating. In early amnion rupture, deformation and disruption of other structures, including craniofacial and limbs, can be demonstrated (limb body wall complex) (Tsirka et al, 2007, Russo et al, 1993).

BSA has sometimes been included in the very heterogeneous group of fetal conditions known as limb body wall complex (LBWC); however, many authors consider BSA a separate entity (Plakkal et al, 2008 Kocherla et al, 2015).

The most accepted defini tion of LBWC has been meeting two of the following three criteria: (1) thoraco abdominoschisis or abdominoschisis, (2) limb defects and (3) craniofacial defects such as cleft lip/palate and encephalocele. While with BSA, the cord is absent, the abdominal organs are herniated and attached to the placenta, there is kyphoscoliosis and positional abnormalities of the lower limbs. Many of the anomalies of LBWC are found in amniotic band sequence including the craniofacial and limb defects and the two conditions are considered to overlap (Bianchi et al, 2000, Russo et al, 1993). Most authors consider LBWC

to result from early amnion rupture (Tsirka et al, 2007, Russo et al, 1993), while BSA is thought to be a consequence of abnormal embryonic folding. These features help in the ultrasonographic differential diagnosis of BSA and amniotic band sequence/LBWC in the first trimester.

Accurate prenatal diagnosis of BSA is feasible from at least as early as the 11th week of pregnancy and the condition can be easily distinguished from other abdominal wall defects. Early accurate diagnosis allows families reproductive choice and optimal counselling about postnatal outcome.

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 . Defects in genes related to embryogenesis may play a role (Kocherla et al, 2015).

The germinal disk abnormality is thought to represent complete failure of body folding along all three axes (cephalic, caudal, and lateral) (Plakkal et al, 2008). Normal body folding results in separation of the intra embryonic coelom (future peritoneal cavity) from the extra embryonic coelom, formation of the body stalk, and development of the umbilical cord (Tsirka et al, 2007, Bianchi et al, 2000). 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 mid abdominal contents into a large wide based amnio peritoneal sac, which inserts peripherally onto the placental chorionic plate in lieu of an umbilical cord or with a very short umbilical cord (Russo et al, 1993, Plakkal et al, 2008).

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. (Russo et al, 1993, Plakkal et al, 2008).

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 (Kocherla et al, 2015). 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 (Plakkal et al, 2008), each being the consequence of different pathogenic mechanisms (Daskalakis et al, 2015):1.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. 2.The placental-abdominal type in which no craniofacial defects are present, but which involves urogenital anomalies, anal atresia, lumbosacral meningocele, short cord, persistence of extra embryonic 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.

REFERENCES

Daskalakis G, Pilalis A, Papadopoulos D, Antsaklis A. Body stalk anomaly diagnosed in the 2nd trimester. *Fetal Diagn Ther.* 2003;18:342–344.

Tsirka A, Korkontzelos I, Diamantopoulos P, Tsirkas P, Stefos T. Prenatal diagnosis of body stalk anomaly in the first trimester of pregnancy. *J Matern Fetal Neonatal Med.* 2007;20:183–184.

Kähler C, Humbsch K, Schneider U, Seewald HJ. A case report of body stalk anomaly complicating a twin pregnancy. *Arch Gynecol Obstet.* 2003;268:245–247.

Bianchi DW, Crombleholme TM, D'Alton ME. *Fetology: Diagnosis and Management of the Fetal Patient.* New York: McGraw--Hill Professional; 2000. Body-stalk anomaly; p. 453.

Russo R, D'Armiento M, Angrisani P, Vecchione R. Limb body wall complex: A critical review and a nosological proposal. *Am J Med Genet.* 1993;47:893–900.

Plakkal N, John J, Jacob SE, Chithira J, Sampath S. Limb body wall complex in a still born fetus: a case report. *Cases J.* 2008;1:86.

Kocherla K, Kumari V, Kocherla PR. Prenatal diagnosis of body stalk complex: A rare entity and review of literature. *Indian J Radiol Imaging.* 2015;25:67–70.