BODY STALK ANOMALY: CLINICAL AND HISTOPATHOLOGIC FINDINGS OF THIS RARE ANOMALY IN A NIGERIAN NEWBORN

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ABSTRACT

Introduction: **Body stalk anomaly (BSA) is a rare and severe congenital malformation in which the exact pathophysiology is still unknown. The 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.**

Case presentation: **We report a case of sonologically delayed diagnosis of BSA which was confirmed post-delivery following histopathological examination and we reviewed relevant literature regarding this phenomenon. Sonographic features of the foetus included a wide anterior abdominal wall defect (omphalocele) with protrusion of the liver into the amniotic cavity. The umbilical arteries show normal calibre, flow, velocimetry, and spectral waveform.**

Conclusion: **Body stalk anomaly is accepted as a fatal anomaly, so it is important to differentiate it from other anterior abdominal wall defects prenatally and this could guide the management options.**

Keywords: Body stalk anomaly, Anterior abdominal wall defect, Prenatal ultrasound scan, Nigerian

INTRODUCTION

Body Stalk Anomaly (BSA), is a rare foetal congenital anomaly with an almost universally fatal outcome postnatally.[1] The incidence at birth is about 0.2-0.3/ 100,000 live births because most of the affected foetuses undergo intrauterine deaths.[2, 3.]

In a recent multicenter study by Daskalakis et al.[4] in which 106,727 foetuses between 10 and 14 weeks of gestation were analysed, an incidence of 1/7,500 pregnancies were found to be affected. 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. Of all the types of anterior abdominal wall defects, body stalk anomaly is the 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.[4] The presence of the liver and intestine in the extraembryonic coelom differentiates body stalk anomalies from other subtypes. Body stalk anomaly is generally not associated with chromosomal anomalies[4], however it might occur in conjunction with neural tube defects, genitourinary malformations, abnormalities of the chest wall, intestinal atresia, and craniofacial defects, among others.[5,6] 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.[7,8]

CASE PRESENTATION

We report a case of sonologically delayed diagnosis of BSA in a primigravida and this was confirmed post-delivery following histopathological examination.

M. Baby was a 10-minute old neonate delivered to a 26-year-old primigravida at term via an elective Caesarean section. APGAR score at delivery was poor with a birth weight of 1.5 kg. Prior to presentation and subsequent delivery, the mother booked the pregnancy during the first trimester and had routine antenatal care. She had two obstetric scans in the first trimester which did not reveal any anomaly, however a repeat done at another facility in the late second trimester showed a complex anterior abdominal wall defect (Figure 1) necessitating the request by the parents for another repeat at a different facility to confirm the earlier finding. At presentation in our facility for expert

care, an obstetric Doppler ultrasound scan was done and showed a 33 weeks + 2 days fetus with a normally located placenta. There was a wide anterior abdominal wall defect with protrusion of the liver into the amniotic cavity and the amniotic fluid volume was normal. The umbilical arteries showed normal calibre, flow, velocimetry, and spectral waveform. A prenatal diagnosis of Omphalocele major, abdominoschisis and OEIS (Omphalocele, exstrophy of bladder and colon, imperforate anus, and spinal defects) syndrome were considered based on the complexity of the ultrasound findings.

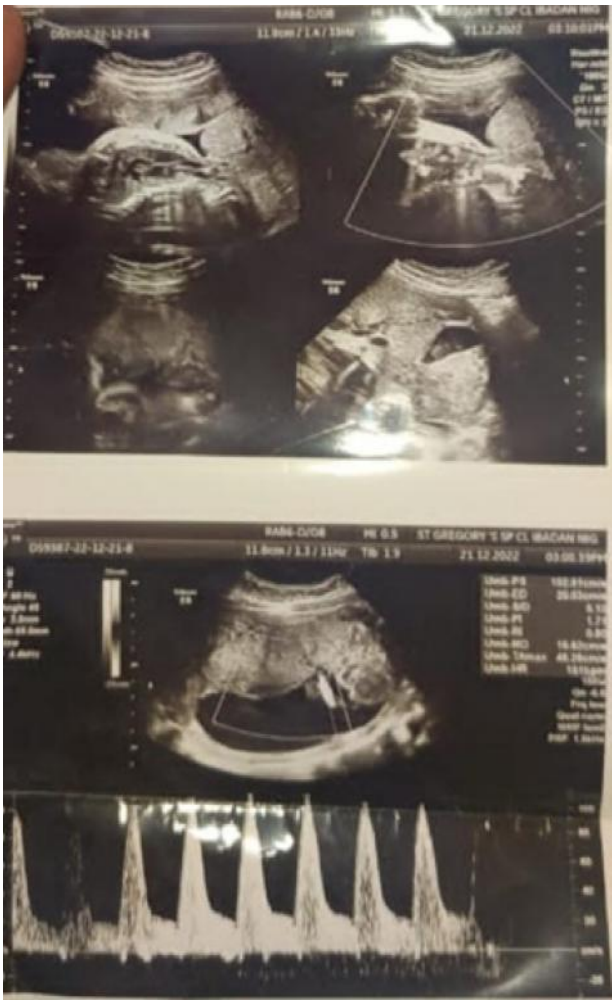

Figure 1: Obstetrics scan image done at 31 weeks with large anterior abdominal wall defect.

At birth, examination showed an inactive newborn, with poor Apgar score and multiple congenital anomalies with the placenta attached. (Figure 2)

The neonate did not survive beyond 15 minutes of birth due to the lethal nature of the anomaly. Findings at autopsy (Figure 3) revealed a neonate with ambiguous external genitalia, the placenta was seen covering a large portion of the anterior abdominal wall defect with a transparent sac. The combined weight

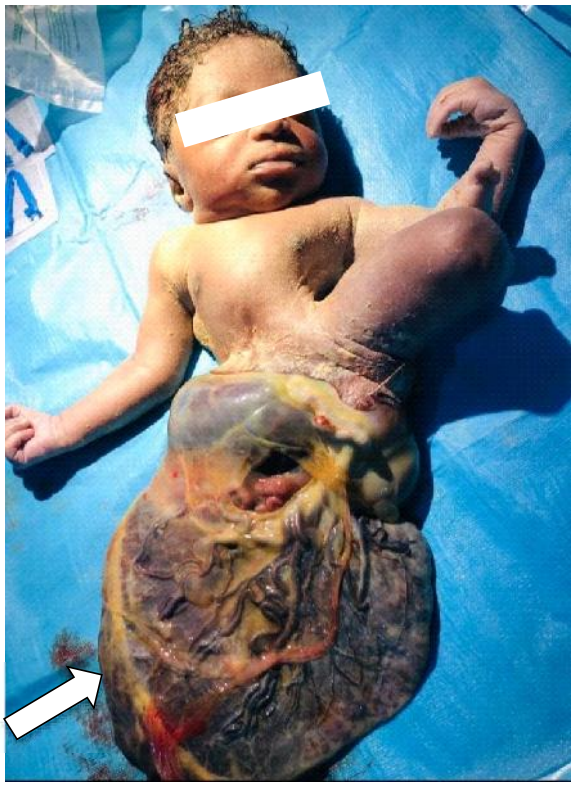

Figure 2: Immediate post-delivery, photograph depicting a body stalk anomaly.
(White arrow: Attached placenta)

of the patient and placenta was 1.59 kg while the placenta alone weighed 390 g. A short umbilical cord was visualised, 1.52 cm in length with three umbilical vessels.

There was a kyphoscoliosis involving the anterior chest wall and spine. The right lower limb was absent, while the left lower limb was deformed with three toes seen on the forefoot (oligodactyly).

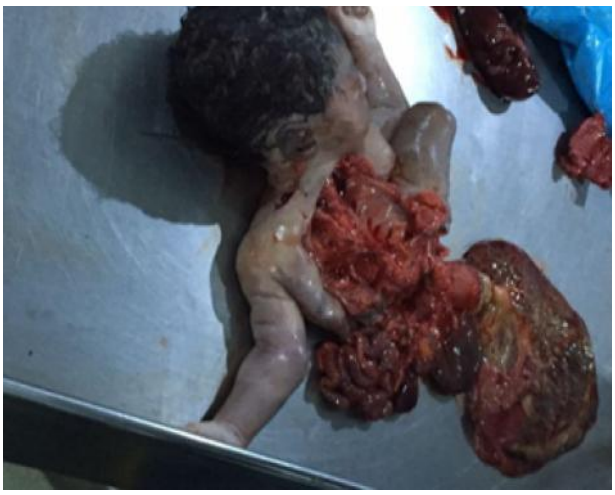

Figure 3: At autopsy to evaluate histopathologic features.

The right and left lungs appeared normal. The right lung had three lobes, and the left lung had two lobes, which were both collapsed with a negative flotation test. The heart was located within the thorax, and the chambers were normal except for an atrial septal defect seen.

Both kidneys and ureters were seen within the abdominal cavity and appeared normal. There is an accessory spleen. The external genital appeared immature with an anal opening smeared with meconium.

A diagnosis of a placental-abdominal type of body stalk anomaly (Limb body wall complex), Swanson type I, II, and IV for the left limb anomaly were made post-delivery (Figures 2).

The parents were counselled and reassured as this complex malformation has no genetic or familial predilection because of their worry about future pregnancies.[2]

DISCUSSION

BSA 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 on sonography. Body stalk defects can be detected at the end of the first trimester of pregnancy by ultrasound scan. 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 syndrome.[9] 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 which is thought to represent a complete failure of body folding along all three axes.[10,11] 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.[12,13] 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.[12,13] Defects in genes related to embryogenesis may play a role, however, causality has not been established.[14]

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.[12,13]

Van Allen *et al.*[10] set forth the diagnostic criteria for BSA in 1987. Two of the three following anomalies must be presented to establish a positive diagnosis:[10,15.]

1. Exencephaly/encephalocele with facial clefts
2. Thoracoschisis and abdominoschisis (midline defect)
3. Limb defect (for example club foot, polydactyly, oligodactyly, syndactyly, brachydactyly, amelia).

Two main phenotypes have been described in the literature,[16,17] each being the consequence of different pathogenic mechanisms:[15]

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 rectal anomalies, lumbosacral meningocele, short umbilical cord, persistence of extraembryonic coelom and intact amnion– it seems to be due to intrinsic abnormal embryonic development. This type is consistent with the reported case.

CONCLUSION

BSA is a rare fatal congenital malformation syndrome as seen in this index patient and the ultrasonographic findings were consistent with those reported in the literature.[18] Early antenatal sonographic diagnosis is necessary so that the parents can make an informed decision on the options of continuation or termination of pregnancy. Expertise in prenatal detection of congenital anomalies is invaluable and rewarding during antenatal care period in our setting.

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