Deciphering Body Stalk Anomaly: A Rare Case Presentation and Review

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Abstract

Body stalk anomaly (BSA) is a rare and lethal developmental defect affecting the thorax and abdomen, with a reported prevalence of 1 in 7500 births in 10-14 weeks of gestation as reported in United Kingdom[1]. Often diagnosed by the end of the first trimester, it presents with severe kyphoscoliosis and abnormalities in abdominal and thoracic structures, including herniation of viscera. We present a case of BSA in a 22-year-old primigravida, diagnosed at 18 weeks gestation. The anomaly scan revealed characteristic features such as a large anterior abdominal wall defect and abnormal placement of the liver and umbilical cord. Despite advancements in prenatal screening, BSA remains a diagnostic challenge, requiring differentiation from other abdominal wall defects. Early detection through detailed ultrasound in the first trimester is crucial for timely management, typically involving termination of pregnancy due to its incompatible nature with life. Further research is needed to elucidate its etiology and improve diagnostic accuracy.

Keywords: Body Stalk Anomaly; Lethal; Congenital Malformation; Abdominal Wall Defect; Herniated Viscera; Fetal Kyphoscoliosis.

Introduction

Body stalk anomaly (BSA) is a complex congenital anomaly and a lethal malformation with abdominal and thoracic wall defect. Abdominal and thoracic structures develop in the coelomic cavity. Associated anomalies may include, intestinal malrotation with short or absent umbilical cord, kyphoscoliosis, lower extremities anomalies, neural tube defects, genitourinary malformations, intestinal atresia and various anomalies of the chest wall and craniofacial defects. Notably, BSA is typically not associated with chromosomal abnormalities.[2] Here, we describe a case involving a young woman whose fetus exhibited features of BSA, contributing to the existing body of literature on this condition.

Case Report

A 22 year old young primigravida, with non-consanguineous marriage was referred from peripheral hospital at 18 weeks with anomaly scan showing multiple anomalies. Her first trimester was uneventful. Patient received folic supplementation after diagnosis of pregnancy in first trimester. There was no history of consumption of alcohol, smoking cigarettes or marijuana and drug intake (cocaine use). Dating scan was done at 7 weeks which was corresponding with the gestational age. NT scan and dual marker were not done. Glucose tolerance test was normal. Anomaly scan done at 18 weeks showed a single live intrauterine pregnancy corresponding to 18 weeks 4 days with borderline oligohydramnios. There was a large anterior abdominal wall defect with complete outpouching of all viscera limited by a thin membrane. Among the herniated viscera, liver appeared directly attached to the placenta [Figure 1]. Thoracic cavity was contracted and there was severe kyphoscoliosis. Right kidney was multicystic and dysplastic, left kidney and urinary bladder could not be made out separately. Head, neck, facial structures were normal. All fetal long bones, both hands and feet were visualised and normal. With all these findings, diagnosis of body stalk anomaly was established. The patient was counselled regarding the neonatal outcome and the couple decided for termination of pregnancy. After an informed consent, pregnancy termination was done by medical method. TORCH panel and fetal karyotype could not be performed due to patient's financial constraints.

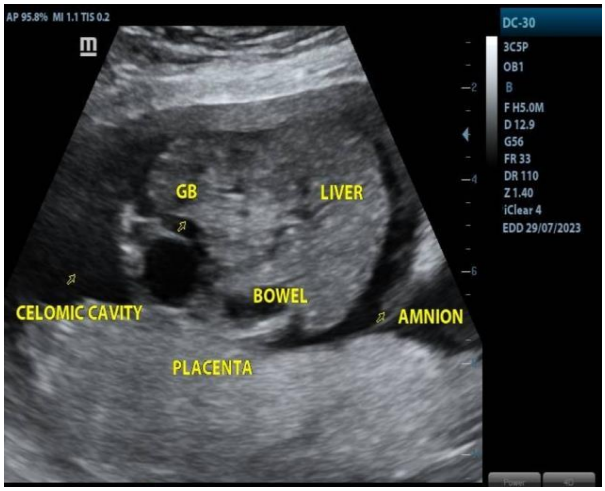

Figure 1: Ultrasonographic picture showing liver and bowel directly floating in the coelomic cavity and liver appears in direct contact with placenta

The aborted fetus showed the following features [Figure 2]: A large abdominal wall defect with herniated viscera containing liver, kidney, intestinal loops covered by amniotic sac. The umbilical cord attached to placenta which was directly attached to the liver. There was severe kyphoscoliosis and the thoracic cavity was contracted, all the four limbs and facial anatomy was normal. Infantogram [Figure 3] confirmed all the skeletal features.

Figure 2: Fetus with large abdominal wall defect, contracted thoracic cavity and placenta with short cord, and all four limbs

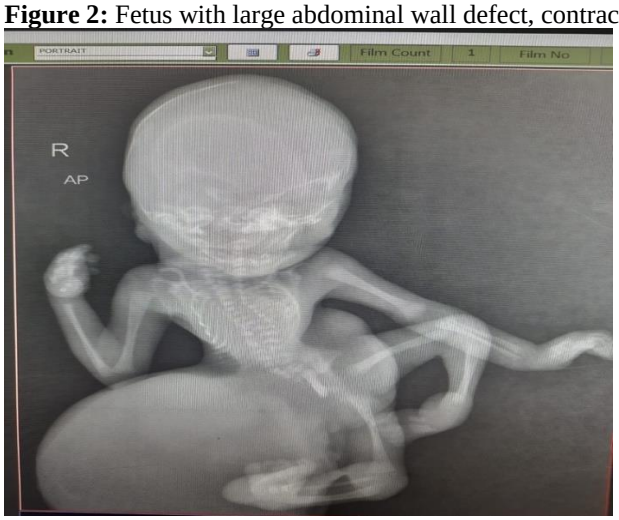

Figure 3: Infantogram of the fetus shows all normal limb bones, scoliosis of thoracic spine and small ribs with contracted thorax

Discussion

BSA is a complex and rare developmental malformation characterised by abdominal or thoracic wall defects, spinal cord and limb deformities. As per study by Routhu M et al, among 32100 pregnant women, referred for routine antenatal scans, ten women were diagnosed to have baby with BSA.[2]

The intrathoracic and abdominal organs lie in the extraembryonic coelom and the viscera usually the liver is directly attached to the placenta or there may be a short umbilical cord. Though the recurrence rate in subsequent pregnancies is very low, the patient should undergo a detailed first trimester anomaly scan as per standard of care.

The exact cause is unknown. Various theories have been proposed like early amnion rupture, vascular disruption and embryonic maldevelopment. Early rupture of the amnion leads to the formation of multiple fibrous bands resulting in structural defects and fetal structures lying outside the amniotic cavity.[3] According to Van Allen[4,5] and colleagues, early generalized compromise of embryonic blood flow during first 4-6 weeks of embryonic development leads to the failure of closure of ventral body wall and persistence of the extra embryonic coelomic cavity. Cocaine use in mother can be responsible for impaired placental perfusion due to vasoconstrictive effect of the drug and subsequent development of BSA. Certain other risk factors have been identified, such as lower socioeconomic status, maternal diabetes and maternal hemorrhagic disorders.[6] In our patient, no such high risk factors were identified.

Russo R[5] and colleagues, noticed two phenotypes of body stalk syndrome. First phenotype is related to early vascular defect leading to craniofacial defects, amniotic bands and/or adhesions. Second variant is attributable to intrinsic embryonic maldevelopment resulting in urogenital anomalies, anal atresia, abdominal placental attachment and persistence of extraembryonic coelom. In our case, there was a large anterior abdominal wall defect with herniation of the abdominal structures. Among the herniated viscera, liver appeared directly attached to the placenta (figure 1). Thoracic cavity was contracted and there was severe kyphoscoliosis. Right kidney was multicystic and dysplastic, left kidney and urinary bladder could not be made out separately. Head, neck, facial structures were normal. All fetal long bones, both hands and feet were visualised and normal. Therefore, our case falls into second variant as there was large abdominal wall defect with persistence of extra embryonic coelom.

The differential diagnosis of body stalk anomaly includes other abdominal wall defects like amniotic band syndrome, gastroschisis, omphalocoele, bladder exstrophy, Pentalogy of Cantrell and cloacal exstrophy.[7,8] It is the presence of the liver and intestine in the extraembryonic coelom that differentiates body stalk anomalies from other abdominal wall defects. Foetus appears adherent to the placenta and there is no free flowing umbilical cord. In omphalocele the cord inserts into the apex of the omphalocele membrane and is usually associated with chromosomal anomalies. In gastroschisis, umbilical cord inserts adjacent to the abdominal wall defect and associated anatomic defects are uncommon. Umbilical hernia is the defect in the linea alba and otherwise intact abdominal wall. In Pentology of Cantrell, cord is inserted into omphalocele membrane and associated abnormalities are cephalad to the umbilical insertion. Ectopia cardis may be present. In bladder exstrophy, cord insertion is lower than the normal on foetal abdomen, abdominal wall defect is below the umbilicus and non-visualization of the bladder is the key finding. In cloacal exstrophy, there are multiple defects including omphalocele, exstrophy of bladder, imperforate anus, spinal defects.

Nagase H et al[9] studied pregnancy outcome in 27 cases of body stalk anomaly. 5 pregnancies resulted in livebirth, mean gestational age at delivery being 33 weeks. All five liveborn neonates died within few hours of birth. Due to lethal nature of this anomaly, patient and relatives need to be well informed about the fetal prognosis and need for termination of the pregnancy. The fetus usually dies shortly after delivery and there are no specific therapeutic interventions. If the patient still wants to continue the pregnancy, vaginal delivery is preferred and caesarean section is indicated only for obstetric indications.

It may be associated with increase in the serum alpha-fetoprotein levels in 100% of the cases mainly in the second trimester of the pregnancy.[10] It should be suspected when a large abdominal defect is observed as well as abnormalities in the axial skeleton such as kyphosis or scoliosis, and a short or absent umbilical cord is present.

According to Argyro Syngelaki et al,[11] a basic ultrasound scan which aims to obtain the appropriate mid-sagittal view of the fetus for measurement of the CRL and NT and transverse sweeps through the head and abdomen should identify all cases of body stalk anomaly, anencephaly, alobar holoprosencephaly, exomphalos, gastroschisis and megacystis.

The main limitation is that this is only a description of a single case. Further research is needed to understand the pathophysiology of this lethal anomaly.

Conclusion

BSA is incompatible with life and hence needs termination of pregnancy in all diagnosed cases. Therefore, it is important to differentiate this anomaly from anterior abdominal wall defects like omphalocele, gastroschisis and bladder exstrophy as these are compatible with life if not associated with chromosomal anomalies. Early detection by a detailed first trimester ultrasound should detect all cases of body stalk anomaly and pregnancy termination can be offered.

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