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A case of body stalk anomaly: the value of adding fetal magnetic resonance imaging to ultrasonography for perinatal management

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Summary

Body stalk anomaly (BSA) is a rare congenital anomaly with an abdominal wall defect and is associated limb and spine abnormalities. As BSA is invariably fatal, it is very important to differentiate BSA from potentially treatable abdominal wall defects, such as gastroschisis. The authors report a case of BSA that was diagnosed prenatally using MRI. On prenatal ultrasonographic examination at 30 weeks of gestation, severe gastroschisis was suspected. However, subsequent MRI clearly demonstrated characteristic findings of BSA. The fetus was delivered at 32 weeks of gestation. The parents did not consent to aggressive treatment and the fetus died a few minutes after birth. Fetal MRI is a powerful adjunct to ultrasonography; it provides more comprehensive images of BSA, enabling prudent perinatal management.

Key words: Body stalk anomaly; Ultrasonography; Magnetic resonance imaging; Fetus.

Introduction

Body stalk anomaly (BSA), a subtype of anterior abdominal wall defect, is characterized by an absent or short umbilical cord with associated severe spinal scoliosis and limb defects [1]. Although the exact etiology of this condition is still unclear, three general pathogenic mechanisms have been proposed for this disorder: amnion rupture, vascular disruption, and embryonic malformation [2]. BSA is a rare malformation syndrome with a reported incidence between one in 14,000 and one in 22,000 pregnancies in large epidemiologic studies [3, 4]. However, others reported an incidence of one in 4,000 to one in 7,500 pregnancies [5, 6]. Most are diagnosed by ultrasonography in the prenatal period, and are not continued until birth. Because BSA is lethal in the majority of cases, some authors question the continuation of pregnancy and postnatal aggressive treatment [1]. Therefore, it is important to diagnose these lesions prenatally and to differentiate them accurately from other anterior abdominal wall defects. The authors report a case of BSA that was diagnosed prenatally using MRI.

Case Report

A 24-year-old, healthy pregnant primigravida was transferred to the present hospital at 30 weeks of gestation because of fetal anomalies. There was no history of consanguinity or any family history of malformations. No maternal use of medication or antepartum illnesses were reported. On ultrasonographic examination, a large abdominal wall defect was detected with herniation of the liver and intestine, and gastroschisis or omphalocele was suspected (Figure 1). However, the umbilical cord could not be seen, and craniofacial and limb anomalies were not definitive. Then, fetal MRI was performed, which clearly showed a short umbilical cord and scoliosis of the spine in addition to evisceration (Figure 2) . Thus, the diagnosis of BSA was confirmed. The MRI showed the fetal lungs to be hypoplastic. It was explained to the parents that the fetus had a malformation that had an extremely high mortality rate, and that the fetus would not avoid serious sequelae, even if it were to survive. The fetus was delivered spontaneously at the gestational age of 32 weeks and six days, weighing 1,640 grams. Apgar score was 1 at one minute. The baby died a few minutes after birth without aggressive treatment, to which the parents did not consent. Postpartum physical examination revealed failure of closure of both thoracic and abdominal walls with ectopia cordis, evisceration of the left lung, and abdominal organs, but normal anus, genitalia, and limbs. The thorax was small and bell-shaped, with U-shaped severe scoliosis of the spine. Abdominal viscera were attached directly to the placenta and were covered by a ruptured amniotic sac. A short, malformed umbilical cord was present, incompletely covered by amnion, and the umbilical vessels were embedded in an amniotic sheet that connected the skin margin of the anterior body wall defect to the placenta (Figure 3). Autopsy confirmed the presence of the aforementioned MRI abnormalities. Other autopsy findings included bilateral hypoplastic lung and normal intrinsic cardiac anatomy.

Discussion

BSA is a lethal condition and aggressive surgical treatment is rarely successful. There are no currently available fetal interventions, and there is nearly universal early postnatal death [7]. In addition, Costa *et al.* recently reported an

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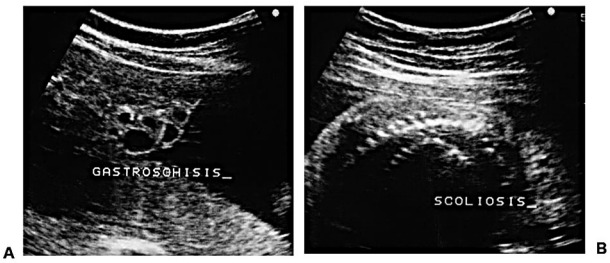

Figure 1. — Sonogram of the fetus shows herniated viscera (A) and scoliosis of the spine (B).

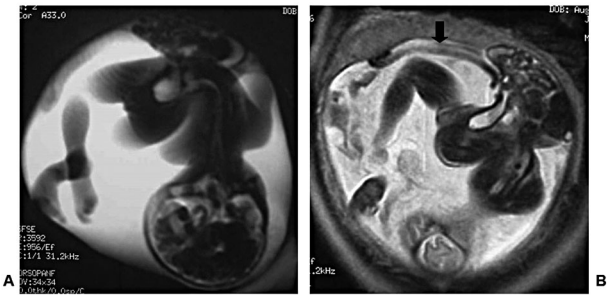

Figure 2. — T2-weighted images obtained by single-shot, fast-spin echo sequences. (A) Most of the viscera lie outside the abdomen. Spinal deformity is apparent. There are no significant abnormalities in the limbs and brain. (B) The umbilical cord can be identified but is very short (arrow).

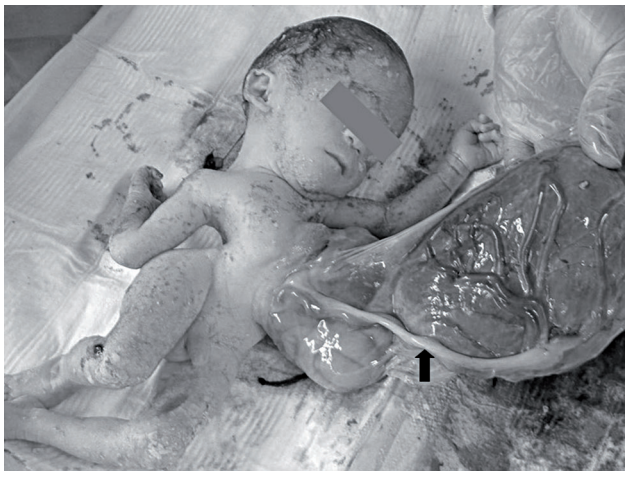

Figure 3. — The newborn demonstrates typical morphologic features of body stalk anomaly, including lateral thoraco-abdominal wall defect and severe scoliosis. The placenta is attached to an extra-embryonic sac containing liver and other herniated viscera. The umbilical cord is malformed and the umbilical vessels are embedded in the amniotic membrane (arrow).

increased risk of maternal morbidity and adverse maternal outcomes in women with late diagnosis or those who choose not to terminate pregnancy [8]. If the pregnancy is continued into the third trimester, complications during labor and cesarean section can occur, including uterine rupture. These points should be discussed during counseling, and termination of pregnancy should be advised when a definitive diagnosis of BSA is made [8]. On the other hand, the differential diagnosis for BSA includes other anterior abdominal and thoracic wall defects, such as omphalocele, gastroschisis, ectopia cordis, pentalogy of Cantrell, and cloacal exstrophy [9]. Among these, gastroschisis is the most benign condition. Therefore, a prenatal diagnosis of BSA and its differentiation from other potentially treatable abdominal wall defects is crucial, as it will influence the choice of treatment and survival. Moreover, long-term survival in BSA has been reported through application of advances in neonatal care [10]. It is important to evaluate the possibility of survival, even after the diagnosis of BSA is confirmed, and it is essential to inform the prospective parents regarding the expectations for infant survival, as well as the outcome and risks and benefits of various approaches to care. Therefore, accurate diagnostic imaging might be required. Ultrasonography (US) is the primary imaging modality for the evaluation of the fetus. It is safe for both fetus and mother, relatively inexpensive, and allows real-time imaging. Several case series and reports of BSA imaged with US have been reported [1, 9], and it is thought that the diagnosis of BSA is usually not difficult to make [11, 12]. However, definitive diagnostic criteria for BSA have not been established because of the variety of its features. The diagnosis of BSA may be difficult sometimes because of the rarity and poor imaging by US, especially in early gestation. In addition, the capability of US to demonstrate fetal abnormalities is limited in cases such as inappropriate fetal position and maternal obesity. In cases of severe oligohydramnios or large maternal body habitus, and in cases in which an unusually complex relationship between the herniated viscera and the deformities exists, the diagnosis of BSA can pose a challenge [13]. In the present case, US did not demonstrate gross craniofacial and limb anomalies, and did not detect an umbilical cord and fetal membranes. Therefore, severe gastroschisis was suspected, and scoliosis of the spine was considered to be a secondary deformity. On the other hand, MRI visualized not only the short umbilical cord but also other characteristic findings of BSA. This case shows that fetal MRI allows visualization of the typical features of BSA. Fetal MRI is a noninvasive, rapid, and highly informative examination that can provide much information on anatomical abnormalities, and the fetal position or mother's physical condition does not affect decision-making. Currently, ultrafast magnetic resonance examinations can produce high-resolution images that are not affected by artifacts of fetal movement, and have been developed for accurate fetal diagnosis [14]. Re-

cently, early prenatal diagnosis of fetal abnormalities using MRI has been reported, particularly for multiple anomalies with poor prognosis [15, 16]. In the present case, MRI findings allowed the parents to have a better visual perception of these conditions. However, only a few reports have provided detailed descriptions regarding the MRI findings in BSA [17]. MRI provides a basis for accurate prenatal diagnosis that facilitates early and informed discussion of the prognosis with the parents. As for safety of fetal MRI, no significant risk to the developing fetus, such as exposure to electromagnetic radiation beyond the first trimester, has been reported [18]. Although US is undoubtedly the first-line diagnostic examination for prenatal congenital anomalies, it may be of value to referring physicians to have an additional diagnostic modality to provide appropriate counseling in difficult situations. MRI may be of great value in the assessment of BSA, and can be complementary to prenatal US. In particular, in utero fast MRI should be considered when fetal US yields ambiguous findings, whether due to patient or technical factors.

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