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Monoamniotic twin pregnancy discordant for body stalk anomaly: a case report

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Abstract

Body stalk anomaly, also known as limb-body syndrome, is a rare and lethal congenital anomaly. It presents a major abdominal wall defect and severe kyphosis with either a rudimentary or absent umbilical cord. We present a case of monoamniotic twin discordant for body stalk anomaly diagnosed at 27 weeks. Ultrasound findings demonstrated a normal Twin A, but Twin B was abnormal with a large ventral wall defect and severe kyphoscoliosis. The pregnancy was managed conservatively and concluded with an emergency caesarean section at 31 weeks on account of preterm labour. The unaffected twin survived with no significant morbidity. This case highlights some of the challenges posed to the obstetrician in managing such a rare anomaly in twin pregnancy.

Keywords: Body stalk, anomaly, monochorionic, monoamniotic pregnancy, foetal ventral wall defect

INTRODUCTION

Body Stalk Anomaly (BSA), also referred to as Limb-Body Wall Complex, is a rare foetal congenital anomaly with an almost universally fatal outcome postnatally. The high fatality is due to the associated pulmonary hypoplasia and other debilitating deformities [1]. There are only a few documented cases of surviving neonates [2]. The anomaly presents a large abdominal wall defect and severe kyphoscoliosis with either a short or rudimentary umbilical cord [3]. The incidence ranges from 1 per 14,000 to 1 per 31,000 pregnancies [4,5]. For example, a multi-centre study by Daskalakis et al. reported an incidence of 1 per 7500 pregnancies based on first-trimester ultrasound prenatal diagnosis [6]. However, the incidence at birth is much lower at 0.2 per 100,000 births [5] because of the high proportion of spontaneous miscarriages in pregnancies with this anomaly [3,7]. Body Stalk Anomaly though rare has a relatively higher incidence in twin pregnancies compared to other abdominal wall defects [10]. A few reported cases of BSA in triplets have also been reported [1,8]. We present a case of a monochorionic-mo Monoamniotic (MCMA) twin pregnancy discordant for BSA that was managed at a Tertiary facility in Ghana.

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CASE PRESENTATION

A 24-year-old Gravida 3 Para 0 with a history of two previous miscarriages at 27 weeks was referred to our tertiary centre on account of MCMA gestation with a gross structural anomaly in the second twin. She was a regular antenatal attendant, had no history of illicit drug use, no family history of congenital anomalies, and no history of consanguinity. The results of her laboratory tests during the antenatal period were unremarkable, and she had no known chronic medical condition. She had a first-trimester ultrasound done at the eight week that reported twin gestations and no other abnormalities. Her second ultrasound at 27 weeks identified the abnormalities that required referral. A detailed ultrasound assessment at our Foetal Assessment centre confirmed an MCMA twin pregnancy with a grossly normal Twin A. Twin B demonstrated a large ventral wall defect and severe kyphoscoliosis (Plate 1A). The liver and intestines, though free-floating, were covered in a membrane adjacent to the placenta (Plate 1B). No free loop of the cord was seen, but umbilical vessels were seen with the colour doppler running from the placenta to the foetus marginally to the eviscerated abdominal content. A diagnosis of

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Monochorionic Monoamniotic twin discordant for Body Stalk Anomaly was made. The mother was counselled on the condition and opted for conservative management. We could not offer selective feticide at our facility because of the advanced gestational age and associated high risk of foetal loss of the normal twin. Instead, foetal surveillance was instituted, comprising twice-weekly non-stress test (NST), foetal dopplers, and twice-daily foetal heart monitoring. At 28 weeks, dexamethasone was given for foetal lung maturity. The goal was to deliver between 32 and 34 weeks gestation. However, the patient went into preterm labour at 31 weeks and had an emergency caesarean delivery. Magnesium sulphate for neuroprotection was given a few hours prior to delivery. Twin A was grossly normal, weighing 1600g with APGARs of 8 and 9 at the first and fifth minutes. Twin B (BSA) died immediately after birth. Post-mortem gross examination confirmed the ultrasonographic findings. The foetus had a large abdominal defect. Significant lordosis and scoliosis were noted. The placenta was attached to the herniated viscera, covered by a thin transparent membrane which was directly continuous with the amniotic membrane of the placenta without any sharp demarcation (Plate 2). The liver, small and large bowels, spleen and

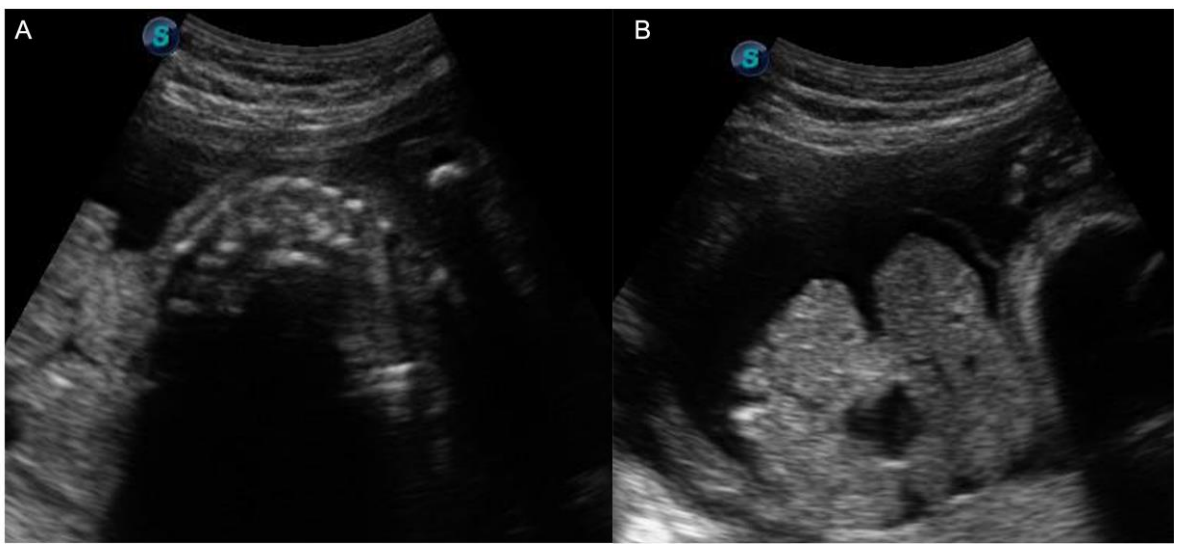

Plate 1: Ultrasound features of BSA Twin

A- Longitudinal section of foetal spine demonstrating severe scoliosis
B- Large abdominal wall defect covered by foetal membranes

Plate 2: Macroscopic examination of the foetus with BSA

A: demonstrates a severely kyphoscoliotic spine and malrotation of the lower limbs
B: demonstrates the large anterior abdomen wall defect with the viscera outside the abdominal cavity covered by a thin membrane

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stomach, were included in the sac. The external genitalia appeared like a female, and the anus was not visible. The pelvis, lumbar spine and upper and lower limbs were grossly distorted. No neural tube defect was noted. Post-mortem radiography showed severe kyphoscoliosis of the thoracolumbar spine with hypoplasia of the thoracic cage, confirming the antenatal ultrasound findings (Plate 3). No abnormalities were found on pathological examination of the placenta and the membranes. The normal twin was sent to the neonatal intensive care unit for admission and assessment. The baby was discharged on day 2 to the mother. The mother was counselled for contraception or preconception care and an early antenatal scan in her next pregnancy.

DISCUSSION

There is an increased risk of foetal anomalies in multiple pregnancies. Compared with singleton pregnancies, monozygotic pregnancies have a higher incidence of ventral body syndromes [13]. However, BSA is rare in monozygotic twins and even rarer in monoamniotic twins [9]. Our case report adds to the few cases documented in literature where most of the monozygotic twins are discordant for this anomaly [11]. No known definitive pathophysiology thoroughly explains BSA [7]. However, three theories have been proposed: early amnion rupture, circulatory insufficiency early in embryonic life, and abnormal germ disc theory. The rupture of amnion theory postulates that amnion rupture leaves the chorionic surfaces exposed to subsequent amniotic bands that cause traumatic injury to the foetus [13]. The circulatory insufficiency theory suggests that vascular disruptions to the embryo around the fifth week of life lead to loss of existing tissue (and thus failure of ventral wall closure) and persistence of the extraembryonic coelom cavity [7,14]. The resulting amniotic tags, ring constrictions and adhesions caused the manifestations of the anomaly seen [14]. This theory has also found support in the association between maternal cocaine use and the development of BSA [7]. However, our case had no history of substance abuse, such as nicotine or marijuana. Abnormal germ disc theory is the most popular of the three theories. The folding of the flat tri-laminar disk in the fifth week of gestation into a cylindrical embryo was disrupted [7]. Four adjacent body structures (one cephalic, two lateral and one caudal) are responsible for folding. Defective folding or malformation of these structures results in BSA [15,9,7].

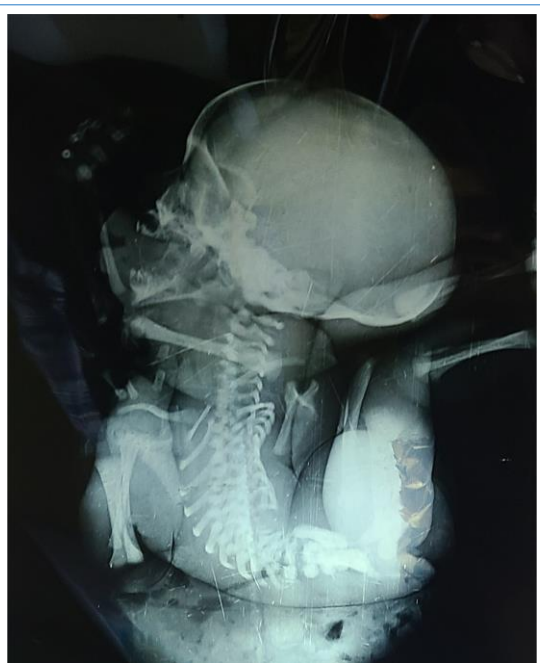

Plate 3: X-ray examination of foetus at delivery. The pelvis and lumberspine were distorted

Two phenotypically distinct groups are described in literature based on the part of the body attached to the placenta. (a) Placenta-cranial is when craniofacial defects (encephalocele, exencephaly) predominate and are seen attached to the placenta by amniotic bands [4,15]. In addition, upper limb defects, as well as thoracoschisis, are common [15]. (b) In the placenta-abdominal phenotypic form, there is the persistence of the extraembryonic coelom, lower limb defects and internal organs defects such as bowel atresia, urogenital abnormalities, short umbilical cord and renal dysplasia (15). The latter accurately describes the case we present in which the foetal liver and bowel were attached to the placenta with a short umbilical cord but no facial defects. Body Stalk Anomaly is a sporadic condition as there is no known underlying genetic cause [8,16]. First-trimester ultrasonography can detect this anomaly even with a 2D grayscale without using a 3D machine [17,4]. This is useful, especially in sub-Saharan Africa, where 2D ultrasounds are more readily available than 3D units.

Ultrasound can serve as a good screening tool and aid in managing such pregnancies as it can afford the mother time for early termination if she so wishes, reducing morbidity and psychosocial stress of terminating a more advanced pregnancy. In our case, this was a monoamniotic twin pregnancy at an advanced gestation on presentation. Unfortunately, many pregnant women in sub-Saharan Africa present for the first antenatal clinic at an advanced gestational age, thus making it difficult for a first-trimester screening. Asah-Opoku et al., in their study, showed that for every ten women who attend an antenatal clinic, only 4 of them would have their first antenatal visit before 16 weeks of gestation [18]. This can be partially resolved with intensive public health education in the communities about the importance of an early clinical and ultrasound assessment at first missed menses. BSA must be differentiated from other ventral wall defects such as gastroschisis, omphalocele and other syndromes with similar presentation such as Pentalogy of Cantrell, Beckwith-Wiedemann Syndrome and OEIS (Omphalocele, exstrophy of cloaca, imperforate anus,

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Table 1: Published case reports of twins with discordant BSA

Reference	Maternal Age (year)	Gestational age at diagnosis (weeks)	Parity	Type of twin Gestation	Management	Gestational age at delivery (weeks)	Outcome
Roxanna E. Bohiltea et al	31	14	G2P1	MCDA	Expectant	33 by C/S	Normal twin A: Female,1.5Kg, APG 7. Abnormal Twin B, died within 30 minutes
Vidaeff et al	26	32	G3P2	MCMA	Expectant	34 by C/S	Normal Twin A:2.870Kg, APG 7, 9 Abnormal Twin B: FSB,1.905
Daskalakis, H.K Nicholaides	20	14	G2P1	MCDA	Expectant	33 by C/S	Normal Twin. Abnormal Twin: 1.55Kg, died within 30 minutes of birth
Daskalakis, H.K Nicholaides	25	14	G1P0	MCMA	Expectant	35 by C/S	Affected baby 1.870Kg, died within 30 minutes of birth
Smrcek et al	29	14	G1P0	MCDA	Expectant	32 by C/S	Abnormal twin died 4 min postpartum; 1360 g; Apgar scores 1/0/0): die
Smrcek et al	34	11	G11P2	TCTA	Feticide of 3rd foetus at 11 weeks	39 by C/S	Healthy male and female
Hirokawa	31	29	G1P0	TC	Expectant	32	Normal twin 1 and 3(weight 1.73 and1.38Kgs resp.) Twin 2 abnormal 1.2Kg, APG 7,5 Died 13 hours after surgery
Kahler et al	33	20	G1P0	DCDA	Expectant	37	Abnormal twin macerated Normal hypotrophic male twin, 2.38Kg

spinal defect) complex [7]. The criteria for diagnosing BSA are finding internal organs in the extraembryonic space with a short/rudimentary or absent umbilical cord on ultrasound [19]. This differs sonographically from an omphalocele, where an umbilical cord inserts directly into the membrane covering the anterior abdominal mass defect [8]. Gastroschisis on ultrasound shows an exteriorized bowel without a covering membrane to the right of a normal umbilical cord insertion [8]. Management of the anomaly in monochorionic twins must be focused on the unaffected twin as complications may arise from the sudden demise of the affected twin or preterm labour, as seen in some cases and ours [13,7]. Intensive antepartum surveillance needs to be instituted for the unaffected twin, and steroids may be given in anticipation of possible preterm delivery. If identified early enough, selective feticide is a plausible option. Bergamelli et al. [12] described the various limitations associated with performing such a procedure in BSA, especially where most cases have a very short umbilical cord. Such cases are likely to require multi-target applications with high-energy delivery close to the placenta with a significant risk of thermal damage. Table 1 summarises the literature search on similar cases and demonstrates that with conservative management, most authors were able to carry the pregnancy to an advanced gestational age with satisfactory outcomes to the normal twin.

Conclusion

BSA is a rare foetal anomaly that presents various challenges to the obstetrician and parents, especially if it occurs in twins with discordance. However, with proper antepartum surveillance and a good neonatal intensive care service, such pregnancies can be allowed to progress. Obstetric ultrasound training should be an integral part of medical personnel training in order to pick up these anomalies early for appropriate referral and management.

DECLARATIONS

Ethical considerations

None

Consent to publish

All authors agreed to the content of the final paper.

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Competing Interests

No potential conflict of interest was reported by the authors.

Author contributions

All the authors (ASD, AKB, JOA, PS, BOF, JC, & IK) contributed to the management of the case, drafting of the report, and final review of the report.

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Availability of data
Data is available upon request to the corresponding author.

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