CASE REPORT

Dichorionic twins discordant for body-stalk anomaly: a management challenge

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SUMMARY

Body-stalk anomaly is a sporadic and rare maldevelopment disorder characterised by large abdominal wall defect, spinal deformity and rudimentary umbilical cord. It is considered a lethal condition as there are only few reports of survival but there was at least one case of long-term survival after neonatal surgery. Differential diagnosis includes isolated omphalocele or gastroschisis, short umbilical cord, amniotic band, limb body-wall complex and other polymalformative syndromes. There are few reports about the expectant prenatal management of the body stalk anomaly as the majority of prenatal diagnosed cases undergo early elective termination. Twin pregnancies discordant for the anomaly represent a challenge to prenatal management as a healthy fetus should also be considered. We describe a case of dichorionic-diamniotic twins discordant for body stalk anomaly which underwent selective feticide of the affected fetus late in pregnancy, in accordance with parents' decision focused on the neonatal well-being of the unaffected twin.

BACKGROUND

Body-stalk anomaly is a sporadic and rare development malformation characterised by a large abdominal wall defect, severe kyphoscoliosis and a rudimentary umbilical cord.^{1,2}

In the first trimester, the prevalence of this disorder is estimated as 1 in 7500 pregnancies, decreasing to 1 in 14 000 births due to the high incidence of spontaneous abortion and elective termination of most of the prenatal diagnosed cases.¹⁻³

The body-stalk anomaly results from a maldevelopment of the cephalic, caudal and lateral embryonic folds during the fifth week of embryogenesis, leading to the absence or shortening of the umbilical cord with the abdominal organs lying outside the abdominal cavity and almost directly attached to the placenta.⁴

There have been several theories on the pathogenesis of this anomaly, including early amnion rupture with direct mechanical pressure and formation of amniotic bands, vascular disruption with compromise of the embryonic blood flow that could lead to a closure failure of the ventral body wall or an abnormality of the germinal disk that leads to a formation of an anomalous amniotic cavity.^{1,3,5-7} There seems to be an association of this anomaly with young maternal age, monozygotic twinning, triplet pregnancy and maternal cocaine abuse.^{4,8,9}

The sonographic findings of the body-stalk anomaly in the first trimester include the demonstration of the abdominal organs in the extraembryonic coelom and an absent or short umbilical cord, usually with a single umbilical artery.¹⁰⁻¹⁴ The diagnosis may be more difficult during the second trimester but a large body wall defect associated with skeletal abnormalities and an absent or very rudimentary umbilical cord, often continuous with the placenta, strongly suggest this condition.¹⁵

The differential diagnosis includes isolated omphalocele or gastroschisis, short umbilical cord syndrome, amniotic band syndrome, limb body-wall (LBW) complex and other polymalformative syndromes.

The body-stalk anomaly has a poor prognosis, being associated with a high incidence of spontaneous abortion and is generally considered to be lethal,^{6,16} although there have been few cases of survival.¹⁷

The recurrence risk in a subsequent pregnancy is low, although there is at least one case of recurrence described.¹⁸

There are only few reports about the expectant management of the body-stalk anomaly as the majority of prenatal diagnosed cases undergo elective termination, but twin pregnancies discordant for the anomaly represent a challenge to the prenatal management as the risk of perinatal morbidity and mortality after iatrogenous preterm delivery of the unaffected infant must be considered.¹⁹

In 2002, Daskalakis and Nicolaides published two case reports of monozygotic twins discordant for the anomaly managed expectantly⁶ and in 2008 Spiller described two similar cases of dizygotic twins. All of them resulted in early neonatal demise of the abnormal twins.³ In 2015, Krishna reported one case of a dichorionic pregnancy discordant for the anomaly where one twin underwent feticide at 18 weeks of gestation, with survival of the other twin.¹⁹

The objective of this article is to describe the ultrasound features and discuss the prenatal management of a dichorionic twins discordant for body-stalk anomaly case. Although we can find some case reports of twins discordant for body-stalk anomaly, we believe our study is the first to manage the pregnancy expectantly until late third trimester, followed by late elective feticide of the affected twin in order to decrease the risk of perinatal morbidity and mortality of the unaffected twin.

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Figure 1 Ultra sound features of a body-stalk anomaly in one twin of a diamniotic-dichorionic pregnancy. At 12 weeks and 3 days of gestation, a large abdominal wall defect covered by fetal membranes (A) was seen in the first fetus, associated with a nuchal translucency above the 99th centile (B). At 19 weeks of gestation, besides the large omphalocele, a fetal spine deformity (C) and a short umbilical cord (D) suggested the diagnosis of a body stalk anomaly.

CASE PRESENTATION

A 26-year-old primigravida was referred to our department for routine first trimester ultrasonography. There was no relevant medical history, usual medication or drug abuse, apart from being a life-long smoker. A detailed ultrasound evaluation revealed a dichorionic-diamniotic twin pregnancy. The crown-rump lengths of both fetuses were correlating to 12 weeks and 3 days of gestation and consistent with the patient's last menstrual period. However, a large abdominal wall defect covered by fetal membranes and with the umbilical cord inserted onto the sac's membrane was seen in the first fetus associated with a nuchal translucency above the 99th centile (figure 1). The second fetus had a normal nuchal translucency and no abnormalities were identified. Due to the presence of sonographic markers suggestive of aneuploidy, a chorionic villus sampling (CVS) was performed at 13 weeks and 4 days of gestation. The karyotype was normal for both fetuses (including the array comparative genomic hybridization testing). At 19 weeks of gestation, an early detailed morphology scan confirmed the presence of a large omphalocele, a fetal spine deformity and also revealed a short umbilical cord on the first twin (figure 1), suggesting a body-stalk anomaly and a morphologically normal second twin with appropriate development.

INVESTIGATIONS

After parents counselling, expectant management was decided due to the presence of a healthy fetus. Serial ultrasound scans were performed. At 30 weeks of gestation, the ultrasound scan revealed a live first twin with appropriate growth but a short umbilical cord, a large omphalocele attached to the placenta, a severe kyphoscoliosis and thoracic hypoplasia. The heart and limbs of the affected twin could never be well evaluated due to the poor image quality conditioned by the fetal position and spinal deformity. The other twin had appropriate growth and amniotic fluid volume.

DIFFERENTIAL DIAGNOSIS

The main syndromes included in the differential diagnosis were Beckwith-Wiedemann syndrome, pentology of Cantrell, cloacal

Figure 2 Gross features showing diamniotic-dichorionic (fused) twin placenta and fetus with desquamation of more 50% of body (maceration) related to retention after fetal medical interruption. (A) Microscopic section of dividing membrane (arrows), inset: (a, amnion; c, chorion). (B) Severe cleft of the abdominal wall (abdominoschisis) without amniotic bands and a very small velamentous umbilical cord it is continuous with the placenta. (C and D) Fetus with lower body wall defect and lower limb defects (cyllosomas) and scoliosis.

exstrophy, amniotic band syndrome, limb body-wall complex (LBW) and short umbilical cord syndrome.

TREATMENT

Outcome and follow-up

At 31 weeks and 3 days of gestation, the patient was admitted to hospital for maternal and fetal monitoring because of an asymptomatic short cervix found at the ultrasound. After prenatal counselling and discussion, it was decided in accordance with parents' decision, to perform selective feticide of the affected twin through cordocentesis. Tocolysis and fetal lung maturity therapy were administered before the procedure. Five days later, the patient went into spontaneous labour and delivered vaginally a stillborn female baby weighing 1710g and a healthy female baby weighing 1920 g. The surviving twin required a vacuum extraction due to prolonged labour. Postmortem evaluation of the stillborn baby confirmed the prenatal diagnosis of body-stalk anomaly with a short umbilical cord with velamentous placental insertion, severe kyphoscoliosis, a large defect of the abdominal wall with evisceration of thoracic, abdominal and retroperitoneal organs covered with fetal membranes (liver, bowel, stomach, spleen, left kidney), pulmonary hypoplasia, bilateral talipes, clinodactyly of the fifth digit of the left hand and dysplastic ears (figure 2).

DISCUSSION

We describe a case of body-stalk anomaly diagnosed on the second trimester of gestation in one of the twins of a dichorionic pregnancy.

Although we can find some case reports of twins discordant for body-stalk anomaly, we believe our study is the first to manage the pregnancy expectantly until late third trimester, followed by late elective feticide of the affected twin in order to decrease the risk of perinatal morbidity and mortality of the unaffected twin.

One can argue that the diagnosis could have been made in the first trimester and managed with a selective feticide with less potential complications at that time, as a large omphalocele and spinal deformity were noticed in the affected twin at 12 weeks of gestation and this anomaly is one of the possible causes for an increased nuchal translucency. Although, a lot of differential diagnosis had to be considered for the nuchal translucency above the 99th centile and associated omphalocele including chromosomal anomalies such as trisomy 21, 13, 18 and triploidies.²⁰ Many polymalformative syndromes such as Beckwith-Wiedemann syndrome (macrosomia, macroglossia, visceromegaly, omphalocele and many other possible associated anomalies),

pentology of Cantrell (thoracoabdominal defects involving the diaphragm, lower stern and pericardium with *ectopia cordis* and large omphalocele) and cloacal exstrophy (omphalocele, exstrophy of bladder and rectum, imperforate anus and spinal defects)^{20 21} can sometimes mimic the spectrum of characteristics of body-stalk anomaly and could not be excluded securely at that time. Also, amniotic band syndrome, LBW complex and short umbilical cord syndrome are probably the diagnosis with more overlap with body-stalk anomaly.

Body-stalk anomaly is characterised by severe cleft of the abdominal wall with rudimental or absent umbilical cord, without amniotic bands. LBW complex, amniotic adhesions and constrictive amniotic bands are discrete but often combined disruption sequences, which some authors consider to correspond to different phenotype manifestations of LBW defects. The LBW complex, although also sporadic and associated with body wall defects with evisceration of thoracic and/or abdominal organs, presents with craniofacial anomalies as exencephaly/encephalocele and facial clefts and is frequently associated with limb defects.^{1 5 22} Amniotic band syndrome and/or amniotic deformities adhesions mutilations (ADAM complex) are also plausible prenatal differential diagnosis characterised by limb malformation and multiple craniofacial, visceral and body wall defects. Some authors even consider the body-stalk anomaly to be a severe presentation of the amniotic band syndrome as amniotic bands are present in 40% of cases moreover related to limb defects.^{14 23} Short umbilical cord syndrome must also be considered in differential diagnosis.

Considering karyotype, the affected twin had a normal karyotype (46, XX), as in the majority of cases reported in the literature, although a case of placental trisomy 16 has been described.^{2 9}

The ultrasound features found in our case were concordant with the previous reports (a large abdominal wall defect, a spinal deformity and a short umbilical cord), although the umbilical cord had all three vessels and not only two, as it is more frequently described in the literature.^{1 3 16}

Considering prognosis, the body-stalk anomaly is associated with a high incidence of spontaneous abortion and is generally considered to be lethal.^{6 16 24} However, some cases of survival have been reported,¹⁷ with at least one case of survival after neonatal surgery with 3 years and 4 months of age of follow-up but with no long-term data available.²⁵ Besides, when the diagnosis is made in the second trimester, as in our case, despite the expected unfavourable prognosis of the affected twin, the risk of a late abortion or early preterm delivery of the healthy twin is a major concern if a selective feticide is performed. The parents should be carefully counselled about their options and the associated risks, namely, the perinatal morbidity and mortality after an early preterm birth. Lipitz *et al* analysed the prognosis of 23 dichorionic twins after performing selective feticide and reported that when the procedure was postponed to 28–33 weeks' gestation, delivery occurred at least 4 weeks later (range 4–11 weeks), with a favourable outcome for the healthy fetus.²⁶ Yaron *et al* also studied 82 twin pregnancies undergoing selective termination and showed that pregnancy loss rate of the procedures performed during the first trimester was 9.7%, compared with 7.8% for terminations performed later in pregnancy.²⁷ Dural *et al* analysed a small series of 31 DC twins and stated that the overall live birth rate after selective feticide was 66.6% in the group of the earliest termination of pregnancy (15–19 weeks) compared with 100% in the groups of later feticide (after 20 weeks), with the rates of extremely and very preterm birth significantly lower for the group in which termination was performed later (30–33 weeks).²⁸

In this case, the parents decided they did not want to risk the rare setting of delivering a live born abnormal twin with limited postnatal survival but they would not also risk undergoing an elective feticide at a gestational age in which the outcome for the healthy twin could be poor. As our national legislation allows pregnancy termination in fatal conditions at any gestational age, it was decided to manage this pregnancy expectantly and discuss the options of keeping this expectant management versus performing an elective feticide closest to the delivery date. However, the patient was admitted at our hospital at 31 weeks of gestation due to a shortening in the cervical length, and although the patient was asymptomatic, this rushed the management discussion and parental decision as a preterm labour could be imminent. After the ethics commission discussion and approval, a selective feticide of the affected twin was performed at 32 weeks of gestation and 5 days later the twins were born. The postmortem study of the affected twin confirmed the diagnosis of body-stalk anomaly.

Learning points

- ▶ A defect of the fetal abdominal wall associated with a rudimental or absent umbilical cord, without amniotic bands, should raise suspicion of a body-stalk anomaly.
- ▶ In cases of dichorionic twins, parents' counselling should be focused not only on the poor prognosis of the affected twin but also on the risks and consequences of an early delivery of the healthy one.
- ▶ In these cases, parents' informed consent should be mandatory.

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