

# Monozygotic twins discordant for body stalk anomaly

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## ABSTRACT

We report on two cases of monozygotic twins discordant for body stalk anomaly, diagnosed prenatally in a multicenter ultrasound screening study at 10–14 weeks of gestation. Ultrasound showed a large abdominal wall defect with most of the abdominal contents and almost half of the body in the celomic cavity, in association with severe kyphoscoliosis and a very short umbilical cord. Both pregnancies were managed expectantly and delivered by Cesarean section. The abnormal babies died soon after birth and autopsy confirmed the sonographic diagnosis. Body stalk anomaly in twins is extremely rare. These are, to our knowledge, the first cases reported on monozygotic twins discordant for this anomaly, indicating that the incomplete twinning theory cannot uniformly explain the pathogenesis of the body stalk in twins.

## INTRODUCTION

Body stalk anomaly is a rare developmental malformation, characterized by an enlarged abdominal wall defect, severe kyphoscoliosis and a rudimentary umbilical cord, with a prevalence of 1 in about 7500 pregnancies in the first trimester<sup>1</sup>. In particular, the presence of this anomaly in monozygotic twins is an extremely unusual condition. To our knowledge, four cases of body stalk anomaly in a twin gestation have been reported so far<sup>2–5</sup> and in only two of them was the diagnosis made by ultrasound; the others were identified by maternal serum alpha-fetoprotein screening in the second trimester.

We report on two cases of body stalk anomaly in monozygotic twins, which were detected in a multicenter ultrasound screening study at 10–14 weeks of gestation.

## CASE REPORTS

### Case 1

A 20-year-old woman, gravida 2 para 1, with a twin pregnancy, was referred to our center at 12 weeks for an abdominal wall anomaly of one of the twins. A detailed ultrasound

evaluation revealed a live diamniotic twin pregnancy with a single placenta, which was classified as monochorionic because of the absence of the lambda sign<sup>6</sup>. Both fetuses were consistent with 14 weeks' gestation and they had normal nuchal translucency thickness measurements. One of them had a large anterior abdominal wall defect with most of the abdominal contents in the celomic cavity. A severe kyphoscoliosis and femur abduction were also observed (Figure 1). These findings suggested a body stalk anomaly. Although this anomaly is lethal, expectant management instead of feticide of the abnormal fetus was decided, for the fear that the prenatal death of the affected fetus could lead to the death of the normal twin. A 20-week scan confirmed the previous diagnosis and revealed inferior deviation of the fetal heart and a sacral spina bifida. Both liver and bowel were eviscerated and the amniotic fluid of the affected fetus was reduced. Normal growth and anatomy of the other fetus were observed. The woman went into spontaneous labor prematurely at 33 weeks and both twins were delivered by emergency Cesarean section. The abnormal neonate weighed



**Figure 1** A longitudinal section of the affected twin demonstrating femur abduction and severe kyphoscoliosis.

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1556 g and died 30 min after birth. There was a single placenta, which weighed 682 g, with two umbilical cords. Postmortem examination confirmed the sonographic diagnosis and a two-vessel cord and a right renal agenesis were revealed in the abnormal twin.

## Case 2

A 25-year-old woman, gravida 1 para 0, was referred to our center at 14 weeks' gestation for the evaluation of a twin pregnancy. One of the fetuses had been diagnosed as having a large exomphalos and abnormal lower limbs. The ultrasound examination demonstrated a live monochorionic monoamniotic twin pregnancy with one normal twin and another with a large anterior abdominal wall defect with evisceration of the liver and bowel, while the lower half of the body was out of the amniotic cavity. A severe kyphoscoliosis and bilateral talipes were also observed. The nuchal translucency thickness was normal in both twins. The diagnosis of a body stalk anomaly in one of the twins was made. Similarly to the first case, expectant management instead of feticide of the abnormal fetus was chosen. Follow-up scans were performed locally and the woman delivered at 35 weeks' gestation by elective Cesarean section. The affected baby weighed 1870 g and died 30 min after its birth. A single placenta with two umbilical cords, weighing 725 g, was delivered. The postmortem confirmed the sonographic diagnosis and revealed a bladder exstrophy. The second twin was normal.

## DISCUSSION

Although many studies of monochorionic twins discordant for several anomalies have been published so far<sup>7,8</sup>, we believe our study to be the first reporting two cases of monozygotic twins with only one fetus affected by body stalk anomaly. Even more interesting is that in both cases the diagnosis was made in the first trimester. One of our cases has already been mentioned in a study reporting a series of monoamniotic twin pregnancies diagnosed at 11–14 weeks' gestation<sup>7</sup>. In four previous cases reported in the literature, the diagnosis was made in the second trimester and both twins were concordant for the anomaly<sup>2–5</sup>. The karyotype of the surviving twins was normal, as in all previous reports of body stalk anomaly.

The sonographic findings of body stalk anomaly in twins at 10–14 weeks were the same as those in singleton pregnancies: a large abdominal wall defect, severe kyphoscoliosis and a very short umbilical cord in one of the fetuses, while the second was normal. In addition, almost half of the body of the affected fetus was in the celomic cavity. Our findings indicate that early amnion rupture before obliteration of the celomic cavity may be the most probable cause of the body stalk anomaly in twins, as in singleton pregnancies<sup>1</sup>. The rupture allows part of the fetal body to pass through the membranes out of the amniotic cavity to the celom, leading to structural defects of the abdominal wall and the spine. Occasional formation of amniotic bands can produce the other associated malformations such as limb amputations, encephalocele and

facial clefts. When this anomaly occurs the fetus has no space in which to move and is almost attached to the placenta. As a consequence the umbilical cord remains very short because its length is related to the tension which is placed upon it by the developing fetus<sup>9,10</sup>.

Alternatively, the abnormal germ disc theory, which is also compatible with the findings in these two cases, has been proposed for the pathogenesis of body stalk anomaly. This theory was first proposed by Streeter<sup>11</sup> and suggests that body stalk anomaly is the result of an abnormal embryonic folding during the 5th gestational week when the trilaminar embryo is transformed into a cylindrical embryo by folding in cephalic, lateral and caudal axes. This leads to a persistence of the celomic cavity, which fails to obliterate, and to abnormal formation of the amniotic sac. A recent report by Paul *et al.*<sup>12</sup> supports this theory, indicating that exocelomic location of the fetus may be one of the features of this anomaly, as the damage occurs before 6 weeks of gestation.

Van Allen *et al.*<sup>13,14</sup> have suggested that early generalized compromise of embryonic blood flow leads to a failure of the ventral body wall to close, with persistence of the celomic cavity. The amnion is then not adequately supported and could be easily ruptured, leading to the formation of amniotic bands. Animal studies, as well as the association of body stalk anomaly with cocaine abuse, a drug with vasoconstrictive properties, have supported this theory<sup>15–17</sup>. A fourth hypothesis for the development of body stalk anomaly in monozygotic twins implicates incomplete division of the single zygote, which leads to a common peritoneal cavity in the pathogenesis of the syndrome<sup>5</sup>. This does not seem to be the cause as is obvious from our report of two cases with only one twin being affected. A female predominance has been mentioned for the body stalk anomaly, as well as for conjoined twins<sup>18,19</sup>. In contrast to this, both our cases concerned male fetuses, an observation which implies that there is no sexual predominance.

The detailed ultrasound assessment of twin pregnancies at 10–14 weeks can easily reveal cases with body stalk anomaly. Proper management with either selective feticide of the affected fetus in dichorionic twins, or expectant management in monochorionic twins, will end in survival of the healthy fetus, while the newborn with the body stalk anomaly is never viable.

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