Ultrasound Obstet Gynecol 2001; **17**: 157–159

A case of body stalk anomaly at 10 weeks of gestation

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KEYWORDS: Body stalk anomaly, Ultrasound, Early pregnancy

ABSTRACT

We describe a case of body stalk anomaly which was diagnosed at 10 weeks of gestation on a dating scan. The fetus was visualized within the exocelomic cavity outside an apparently intact amniotic membrane. The fetus displayed multiple anomalies characteristic of body stalk syndrome including a skull and brain defect, abdominal wall defect, kyphoscoliosis and gross deformities of the lower limbs. These findings do not support early amniotic membrane rupture as the primary event in the pathogenesis of body stalk anomaly and indicate that the exocelomic location of the fetus may actually be one of the features of this complex developmental anomaly.

INTRODUCTION

Body stalk anomaly is a term used to describe a typical pattern of defects that include encephalocele, facial cleft, an anterior abdominal wall defect, kyphoscoliosis, limb deformities and an absent or short monoarterial umbilical cord. The reported prevalence has increased from approximately one in 14 000 pregnancies[1] to one in 7500 in more recent studies[2]. This increase can be explained by more widespread use of ultrasound in the first trimester which enables an accurate diagnosis to be made in cases which are destined to end up in spontaneous miscarriage. A variety of hypotheses have been proposed to explain the pathogenesis of body stalk anomaly. These include embryonic dysplasia[3,4], teratogenic exposure in early pregnancy[5], mechanical damage due to early amnion rupture[6,7] and vascular disruption of the early embryo[8].

We report a case of body stalk anomaly in which the characteristic defects were identified in the first trimester of pregnancy. The features in this case demonstrate that early amnion rupture may not play a role in the etiology of this condition.

CASE REPORT

A 25-year-old primigravid woman with uncertain menstrual dates was referred for an ultrasound scan. She was using the progesterone-only pill for contraception and her cycles were irregular. There was no relevant medical history and she was taking no other medication. Transvaginal ultrasound examination was performed and revealed an intrauterine gestational sac measuring $65 \times 43 \times 40$ mm. The sac contained a live fetus with a crown–rump length of 35 mm, consistent with a gestational age of 10 weeks and 1 day. The amniotic cavity measured $31 \times 31 \times 30$ mm. The amniotic membrane appeared normal and intact (Figure 1). The yolk sac measured $5.3 \times 5.0 \times 4.8$ mm and also appeared normal. The fetus was seen to be developing within the exocelomic cavity and there were multiple fetal abnormalities. The combination of defects comprising a large skull and brain defect, an anterior abdominal wall defect containing liver and bowel, severe kyphoscoliosis and deformed lower limbs was compatible with the diagnosis of body stalk anomaly. The umbilical cord was also located extra-amniotically but appeared to be of normal length (Figures 2–4).

The patient was counseled that this is a lethal condition for the fetus. She chose to undergo surgical termination of pregnancy and an uncomplicated procedure was performed

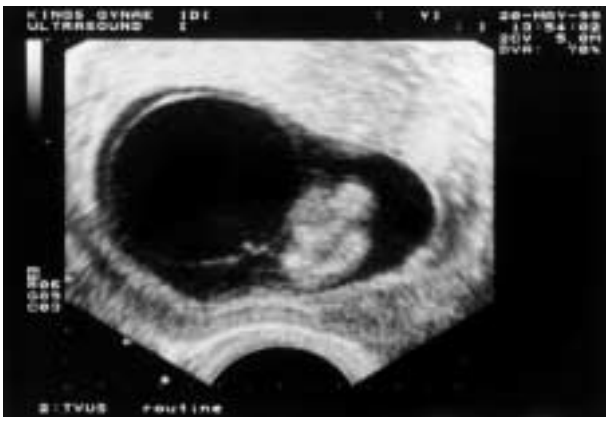

Figure 1 A transverse section of the uterus showing a fetus within the exocelomic cavity (right) adjacent to an intact amniotic membrane.

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Received 23-11-99, Revised 9-8-00, Accepted 2-9-00

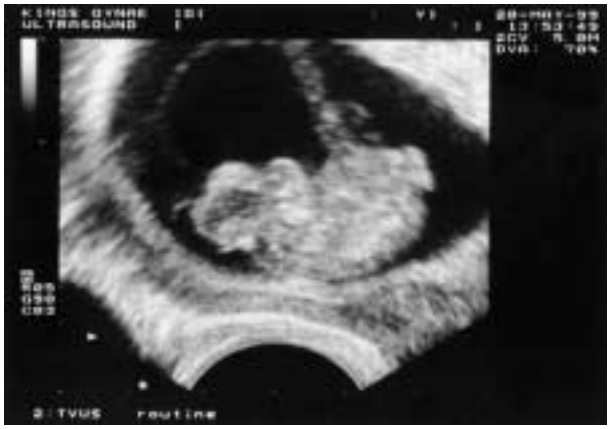

Figure 2 A longitudinal section of the fetus demonstrating a large skull and brain defect and an anterior abdominal wall defect. The umbilical cord is located within the exocelomic cavity but appears morphologically normal.

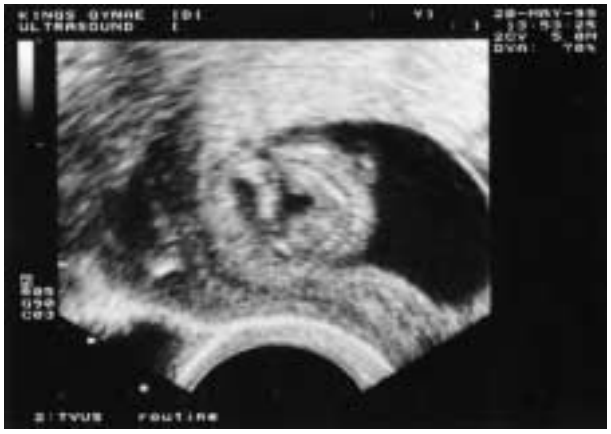

Figure 3 A section through the lumbar spine of the fetus shows severe kyphoscoliosis.

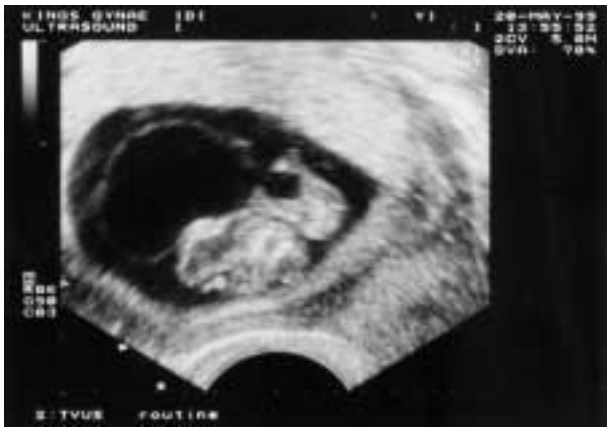

Figure 4 A longitudinal section through the embryo demonstrating abnormal rotation of the lower limbs.

under general anesthetic 2 days later. Unfortunately, the specimen obtained at the surgical termination of pregnancy was severely disrupted which prevented a detailed pathohistologic examination of the fetus.

DISCUSSION

This case demonstrates typical features of body stalk anomaly in a fetus which was developing in the exocelomic cavity. The amniotic cavity was spherical and intact with no signs suggestive of rupture. These findings challenge the validity of the theory that early amniotic rupture with expulsion of the embryo into the exocelomic cavity is responsible for the features of body stalk anomaly. This is in agreement with previous reports on the histopathologic examination of the placenta and membranes which have failed to demonstrate any evidence of amnion rupture in the presence of a fetus displaying the typical features of body stalk anomaly[9]. Others have also described the typical features of body stalk anomaly in the absence of amniotic bands[4,8]. A further challenge to the mechanical theory is the frequent association of defects of the internal organs which cannot easily be explained by the presence of amniotic bands.

Another explanation of body stalk syndrome is the theory of vascular disruption during the first 4–6 weeks of gestation developed by Van Allen *et al.*[8]. The main experimental evidence for this theory was derived from animal studies in which artificial rupture of early amniotic membrane led to major disturbance of rat peripheral vasculature. These changes resulted in similar external defects to those seen in human fetuses affected by the body stalk anomaly. Other studies have shown that alternative causes of vascular disruption such as trauma, hypoxia and use of vasoconstrictive agents in early pregnancy can also lead to similar abnormalities. The finding of an intact amniotic membrane does not uphold the hypothesis of amniotic rupture as a cause of vascular disruption in the present case, neither was there any history of the other alternative causes.

The remaining theory, and that most compatible with the findings in this case, is that of a developmental anomaly. This theory was first proposed by Streeter[3] in 1930. From the detailed examination of postmortem specimens he demonstrated bands of amniotic tissue that were continuous with, rather than adherent to, the defects in the fetus. The defects seen in his series included all the characteristic features of anencephaly, encephalocele, facial clefts, and limb deformities. His hypothesis was that these defects were the result of abnormal folding of the embryo which caused maldevelopment of the amniotic cavity and the germ-disc[3]. In Van Allen *et al.*'s series of 25 cases[8], the amniotic membrane was continuous with the skin of the body wall defect in 85% which is consistent with the theory of developmental anomaly. Abnormal development at the trilaminar stage would also account for the common finding of a single umbilical artery, which is present in over 50% of cases[5,8] as compared to the incidence in the general population of less than 1%[10].

The well-described pattern of abnormalities seen in body stalk anomaly indicates that the damaging event occurs before 6 weeks of gestation[5,8,11]. It has been suggested that

the defects reflect the embryonic proximity of the organs more closely than the vascular distribution, and that vascular disruption is secondary to hypoplasia of the blood vessels in the affected area rather than being the primary etiologic factor[11]. It is also possible that an embryo which is exposed to the celomic fluid would develop abnormally. The celomic fluid composition differs from that of the amniotic fluid[12] and the relative acidosis may exert a teratogenic effect.

In conclusion, the present case illustrates that early amniotic rupture is not an obligatory event in the development of body stalk anomaly. It is therefore possible that anomalies of the amniotic membrane which are often observed in these cases, as well as in fetuses with body limb defect and amniotic band syndrome, are part of the same developmental abnormality that affects the embryo. We agree with Zimmer and Bronshtein[13] who suggested that all these anomalies are probably different expressions of the midline disruption syndrome.

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