Ultrasound Obstet. Gynecol. 10 (1997) 416–418

Body stalk anomaly at 10–14 weeks of gestation

G. Daskalakis, N. J. Sebire, D. Jurkovic, R. J. M. Snijders and K. H. Nicolaides

Harris Birthright Research Centre for Fetal Medicine, King's College Hospital Medical School, London, UK

Key words: BODY STALK ANOMALY, PRENATAL DIAGNOSIS, ULTRASOUND, FIRST TRIMESTER

ABSTRACT

In a multicenter project of screening for chromosomal defects by fetal nuchal translucency thickness and maternal age at 10–14 weeks, 14 of 106 727 fetuses examined had body stalk anomaly. The ultrasonographic features were a major abdominal wall defect, severe kyphoscoliosis and a short umbilical cord. In all cases, the upper part of the fetal body was in the amniotic cavity, whereas the lower part was in the celomic cavity. The nuchal translucency thickness was above the 95th centile in ten (71.4%) of the cases, but the fetal karyotype was normal in all 12 fetuses evaluated. The findings suggest that early amnion rupture before obliteration of the celomic cavity is a possible cause of the syndrome.

INTRODUCTION

Body stalk anomaly, characterized by the presence of a major abdominal wall defect, severe kyphoscoliosis and a rudimentary umbilical cord, is reported in about 1 per 14 000 births[1]. The pathogenesis of this lethal syndrome is uncertain but possible causes include abnormal folding when the trilaminar embryo is transformed into a cylindrical embryo during the first 4 weeks of development, early amnion rupture with amniotic band syndrome, and early generalized compromise of embryonic blood flow[2–7].

The study reports the prevalence and ultrasound features of body stalk anomaly in a multicenter ultrasound screening study at 10–14 weeks of gestation.

MATERIAL AND METHODS

As part of an ongoing multicenter ultrasound study, started in September 1992 and co-ordinated by the Fetal Medicine Foundation, women in London and the surrounding areas are offered screening for chromosomal defects by a combination of maternal age and fetal nuchal translucency thickness at 10–14 weeks of gestation[8]. The pregnancy is examined for the number of live fetuses, crown–rump length (CRL), nuchal translucency thickness and major defects, such as anencephaly and exomphalos. Ultrasound examinations are performed transabdominaly using a 5-MHz, or a 3.75-MHz curvilinear transducer but, in about 5% of the cases, visualization is poor and a vaginal scan is carried out. Demographic details and ultrasound findings are entered into a computer database at the time of the scans and pregnancy outcome is obtained from the patients themselves or the referring hospitals.

A computer search of the database was carried out to identify all singleton pregnancies with live fetuses at the 10–14-week scan and an estimated date of delivery before 1st August 1997. The database was also searched to identify all cases with a body stalk anomaly.

RESULTS

During the study period, there were 106 727 singleton pregnancies with live fetuses at the 10–14-week scan. The median maternal age was 31 years (range 14–49 years) and the median gestational age was 12 weeks (range 10–14 weeks). Body stalk anomaly was diagnosed in 14 cases (prevalence 1 in about 7500 pregnancies) and the median maternal age was 30 years (range 18–41 years) and the median gestation was 12 weeks (range 10–14 weeks). In all cases, there was kyphoscoliosis and a large abdominal wall defect with evisceration of both the liver and bowel. Furthermore, in all cases, the upper part of the fetal body was in the amniotic cavity, whereas the lower part together with a short umbilical cord were in the celomic cavity (Figure 1). The nuchal translucency thickness was above the 95th centile of the normal range for crown–rump length in ten (71.4%) of the cases. The parents were counselled as to the condition of the fetus and they all elected for termination of pregnancy which was carried out surgically. Fetal karyotyping by cytogenetic examination of chorionic villi was carried out in 12 of the cases and this was normal male in nine cases and normal female in three.

Correspondence: Professor K. H. Nicolaides, Harris Birthright Research Centre for Fetal Medicine, King's College Hospital Medical School, Denmark Hill, London SE5 8RX, UK

Figure 1 Fetus at 12 weeks with body stalk anomaly. The upper part of the body is in the amniotic cavity with reduced amniotic fluid, whereas the lower part is in the celomic cavity

DISCUSSION

This ultrasound screening study has demonstrated that the prevalence of body stalk anomaly at 10–14 weeks of pregnancy is about one in 7500. In a previous retrospective study in Scotland, involving examination of birth records from 142 130 pregnancies, there were ten cases of body stalk anomalies (prevalence of about one in 14 000) and they were all identified by maternal serum α-fetoprotein screening in the second trimester of pregnancy[1]. This apparent discrepancy in prevalence at 10–14 weeks and the second trimester suggests that body stalk anomaly is associated with a high incidence of spontaneous abortion early in the second trimester of pregnancy. In this study, as well as in all previous reports on body stalk anomalies, the fetal karyotype was normal. In contrast, with exomphalos in the first trimester of pregnancy, the prevalence of chromosomal abnormalities, predominantly trisomy 18, is more than 50%[9].

The ultrasonographic findings of body stalk anomaly at 10–14 weeks were those of a large anterior abdominal wall defect, severe kyphoscoliosis and very short umbilical cord, as previously described in the second trimester[2,10]. Additionally, there was evidence of amnion rupture with the upper half of the fetal body being in the amniotic cavity and the lower half in the celomic cavity. On the basis of our findings, it could be postulated that the underlying mechanism for body stalk anomaly is early amnion rupture before obliteration of the celomic cavity. Body stalk anomaly could represent one end of the spectrum of amniotic band syndrome, with the abdominal wall and spinal defects being the mere consequence of passage of the lower half of the fetal body into the celomic cavity through the defect in the amniotic sac. The fetus has no place to move and remains almost attached to the placenta. Limb amputations and encephalocele could be the consequence of entrapment in the celomic cavity of individual fetal limbs and the skull, respectively. Animal and human studies suggest that umbilical cord growth occurs in response to tensile forces placed upon it and relates to the fluid space and movement of the fetus; any situation that limits either space or movement can decrease stretch and consequently length of the cord[11,12].

An alternative hypothesis for the pathogenesis of body stalk anomaly, which is also compatible with our sonographic findings, is abnormal embryonic folding. During the 5th week of gestation, the trilaminar embryo, which is connected to the developing placenta by a body stalk, is transformed into a cylindrical fetus by folding in cephalic, lateral and caudal axes[13]. Body stalk anomaly is due to faulty folding in all three axes and this is also associated with failure of obliteration of the coelomic cavity and abnormal formation of the amniotic sac[2,14–16]. A third hypothesis for the development of body stalk anomaly is early generalized compromise of embryonic blood flow, leading to failure of closure of the ventral body wall and persistence of the celomic cavity[6,7]. This could, in turn, lead to rupture of the amnion which is not now adequately supported and in the formation of amniotic bands. Body stalk anomalies have been associated with cocaine abuse and the underlying mechanism is thought to be impaired placental perfusion due to vasoconstrictive properties of the drug[17–19]. However, none of our patients was under any vasoactive medication.

At the 10–14-week scan, it is important to distinguish exomphalos from body stalk anomaly. Although exomphalos is associated with a high incidence of chromosomal abnormalities, in the chromosomally normal group the prognosis is generally good with survival in more than 80% of cases. In contrast, body stalk anomaly is not associated with chromosomal abnormalities but is always lethal.

ACKNOWLEDGEMENT

This study was supported by a grant from the Fetal Medicine Foundation (Charity No: 1037116).

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