A case of body stalk anomaly at 12 weeks of gestation

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Summary

We report a case of body stalk anomaly which was diagnosed at 12 weeks of gestation on a first trimester scan. The fetus displayed multiple anomalies characteristic of body stalk syndrome including abdominal wall defect, kyphoscoliosis, deformities of the lower limbs and a single umbilical artery. Body stalk anomaly is a rare congenital anomaly with a series of similar clinical manifestations and poor prognosis. The first trimester scan can estimate the risk for chromosomal abnormalities and may also reveal major congenital abnormalities.

Key words: Body stalk anomaly; Ultrasound; Early pregnancy; First trimester scan.

Introduction

Body stalk anomaly (BSA) or limb-body wall complex (LBWC) is a severe congenital anomaly with a series of similar clinical manifestations and poor prognosis [1]. It describes 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. Its prevalence has increased from approximately one in 14,000 pregnancies [2] to one in 7,500 in recent studies [3]. This increase could be explained by the widespread use of first trimester ultrasound which enables a diagnosis to be made in cases which were destined to end up in spontaneous miscarriage [4]. A variety of hypotheses have been proposed to explain the pathogenesis of BSA, including mechanical damage due to early amnion rupture, vascular disruption to the early embryo and teratogenic exposure in early pregnancy. An early amnion rupture before obliteration of the coelomic cavity has been discussed to be the major cause of this anomaly. The extra embryonic coelom fails to obliterate and parts of the fetal body remain in an exocoelomic situation. The intraabdominal content persists in the extra-embryonic coelom in a sac covered by amnion and placenta [3]. Van Allen *et al.* [1] found the anomalies to be secondary to vascular disruption at an early stage of gestation. In this context, an association of BSA to maternal drug abuse, especially cocaine has been described [5].

We report a case of body stalk anomaly which was diagnosed at 12 weeks of gestation on a first trimester scan.

Case Report

A 37-year-old primigravid woman at 11+6 weeks of gestation was referred for the first trimester scan. She had had an episode of bleeding from subchorionic hematoma during the 9th week of gestation (Figure 1) and was taking natural microionized progesterone at a dose of 300 mg daily/po. There was no relevant medical history and she was taking no other medication.

Ultrasound examination revealed an intrauterine gestational sac. The sac contained a live fetus with a crown-rump length (CRL) of 47.3 mm, consistent with a gestational age of 11 weeks and 4 days (Figure 2). The nuchal translucency was normal (2.1 mm). The amniotic membrane appeared normal and intact. The fetus was seen to be in the exocelomic cavity and there were multiple abnormalities. The combination of defects comprising an anterior abdominal wall defect containing liver and bowel, kyphoscoliosis, deformities of the lower limbs and a single umbilical artery (Figure 3), were in accordance with the diagnosis of body stalk anomaly.

The parents were counseled that this is a lethal condition for the fetus. They chose a surgical termination of the pregnancy, so an uncomplicated procedure was performed under general anesthesia the next day. The specimen which was obtained at the procedure was disrupted and a detailed pathologic examination was impossible.

Discussion

A heterogeneous group of congenital defects has been referred to as BSA or LBWC. This case demonstrates typical features of body stalk anomaly in a fetus which was developing in the exocelomic cavity. These findings are in accordance with the theory that early amniotic rupture with expulsion of the embryo into the exocelomic cavity is responsible for the features of BSA. The rupture allows part of the fetal body to pass to the coelomic cavity leading to structural defects, mainly of the abdominal wall and spine. The formation of amniotic bands can produce limb deformities. Abnormal development at the trilaminar stage would account for the common finding of a single umbilical artery, which is present in over 50% [1] of the cases, while in the general population it is less than 1%.

Many authors have discussed possible etiologies of these anomalies including teratogens [6], an amniotic band or rupture sequence [7, 8], intrauterine compression [7], failure in morphogenesis [8], a vascular defect [1], placental insufficiency [9] and multifactorial etiology [10].

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Fig. 1

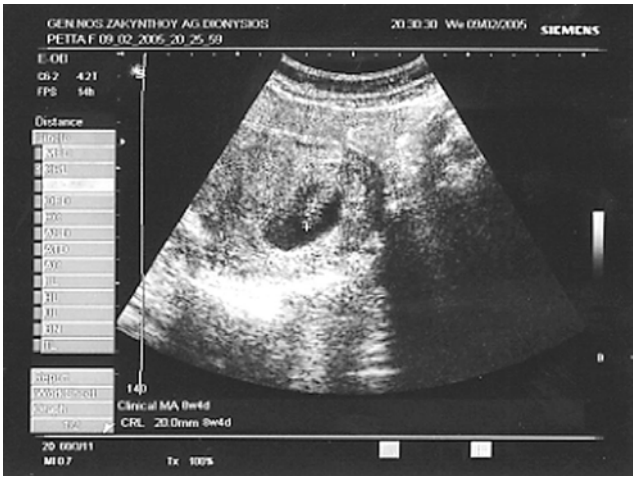

Fig. 2

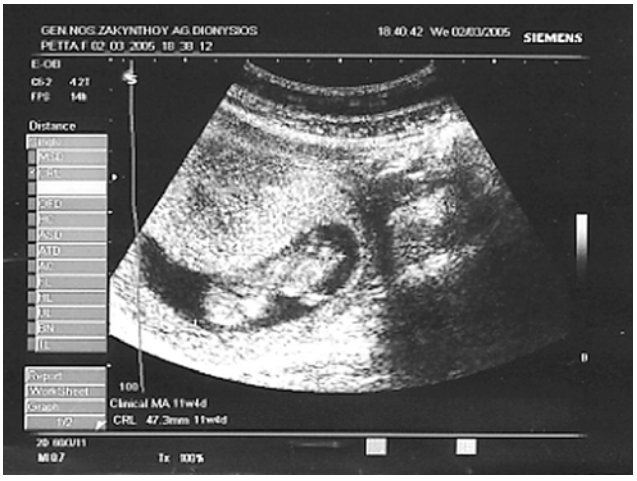

Fig. 3

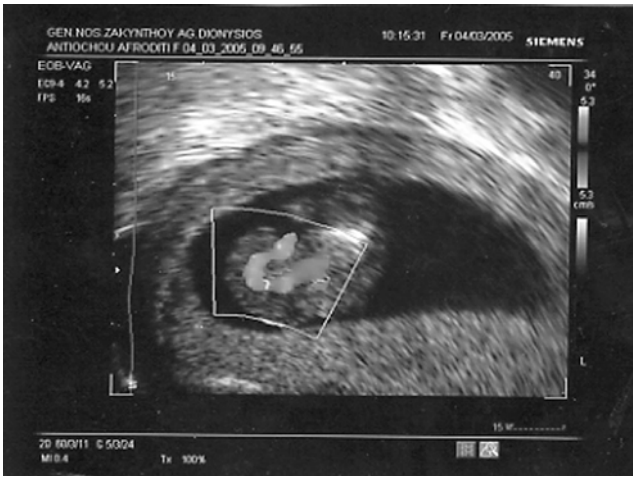

Figure 1. — 9th week scan with subchorionic hematoma (right).

Figure 2. — 1st trimester scan.

Figure 3. — Single umbilical artery.

An explanation of BSA is the theory of vascular disruption during the first four to six weeks of gestation developed by Van Allen *et al*. [1]. They have defined the concept of LBWC as a combination of at least two of the following characteristics: 1) Exencephaly or encephalocele with/without facial clefts, 2) thoracic and/or abdominal ventral body wall defects, and 3)limb defects. Colpaert *et al.* [10] have argued that at least two different types of LBWC can be distinguished from each other. The craniofascial defect type (LBWC type 1), is characterized by abnormal attachment of amnion, which may cause cranial or fascial disruption and/or exencephaly or encephalocele combined with limb defects and/or abdominal wall defects. It is well-explained by an amniotic band or rupture sequence, or a compression theory. The ventral body wall defect type, BSA type or LBWC type 2, is characterized by an umbilical cord anomaly (short, single umbilical artery), thoracic and/or abdominal ventral wall defects with or without limb defects and scoliosis, but does not present with craniofascial defects.

BSA is one of the possible causes when an increased nuchal translucency is observed during the 1st trimester scan [3, 11]. In our case, no increased nuchal translucency was found.

As the condition is practically uniformly lethal to the fetus, as soon as the diagnosis is confirmed, termination in single pregnancies is recommended [2]. In the case of a twin pregnancy with one affected fetus, the proper management consists of selective feticide of the affected one in dichorionic twins, or expectant management in monochorionic twins, so the pregnancy will end in survival of the healthy fetus, while the newborn with BSA is never viable [12].

In conclusion obstetricians, neonatologists and pediatric surgeons must recognize these malformations as lethal and counsel parents to make an informed decision regarding the pregnancy.

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