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Prenatal Diagnosis of Body Stalk Anomaly: A Case Report+

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Body stalk anomaly is a rare developmental anomaly characterized by an enlarged abdominal wall defect, severe kyphoscoliosis and a rudimentary umbilical cord. It is often complicated by anomalies of head, face and extremities. It is diagnosed by ultrasonography, mostly in the second trimester of pregnancy during gestational age dating or evaluation of an elevated maternal serum α-fetoprotein. We present a case of body stalk anomaly at 23 weeks of gestation, diagnosed incidentally during sonographic evaluation in the first visit. Sonographic features of the fetus were severe midline defect of the fetal abdominal wall and a large extra-abdominal mass containing bowel and liver inside, severe scoliosis, clubfoot, absent urinary bladder and oligohydramnios. Body stalk anomaly is accepted as a fatal anomaly, so it's important to differentiate it from other anterior wall defects for evaluating the management options.

Key Words: Body stalk anomaly, prenatal diagnosis

Body Stalk Anomalinin Prenatal Tanısı: Vaka Takdimi

"Body Stalk Anomali" geniş karın ön duvarı defekti, ciddi kifoskolyoz ve rudimenter umblikal kord ile karakterize, nadir görülen gelişimsel bir anomalidir. Genellikle baş, yüz ve ekstremite anomalileri eşlik eder. Sıklıkla gebeliğin ikinci trimesterinde, gestasyonel yaş tayini ya da artmış maternal serum alfa-fetoprotein araştırması esnasında ultrasonografi ile tanı koyulur. Yazımızda gebeliğin 23. haftasında, ilk vizitte ultrasonografi ile tesadüfen tanı koyulmuş body stalk anomalisi vakası sunulmaktadır. Vakanın ultrasonografik bulguları fetusta karın ön duvarında ciddi orta hat defekti ve içerisinde karaciğer ve barsak bulunan abdomen dışında geniş kitle, ciddi skolyoz, clubfoot, idrar torbasının olmaması ve oligohidramniyozdur. Body stalk anomali ölümcül olarak kabul edilmektedir ve tedavi planının yapılabilmesi için diğer karın ön duvarı defektlerinden ayırt edilmelidir.

Anahtar Kelimeler: Body stalk anomali, prenatal tanı.

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Anterior wall defects are characterized into three basic types: omphalocele, gastroschisis, and body stalk anomaly. They occur infrequently and the prognosis for each anomaly is different. Body stalk anomaly also known as Limb Body Wall Complex (LBWC) is the rarest and invariably fatal developmental malformation, characterized by an enlarged abdominal wall defect, severe kyphoscoliosis and a rudimentary umbilical cord. It's often complicated by anomalies of head, face and extremities,[1, 2] They comprise a broad spectrum of phenotypes. Colpaert at al. proposed that body stalk anomaly be classified into two types: type I, complicated by craniofacial defects, and type II, complicated by ventral wall defects without craniofacial defects.[3]

A multicenter study in London and the surrounding areas demonstrated a prevalence of about 1 in 7500 fetuses at 10-14 weeks of gestation.[4] In a retrospective study, involving examination of birth records of 246.189 fetuses delivered alive or dead in Hawaii between 1986-1997, there were 8 cases of body stalk anomaly, with a prevalence of 0.32 per 10.000 live births and fetal death.[5] 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.

We report a case of body stalk anomaly diagnosed in the second trimester, during ultrasound carried out for gestational age dating.

CASE REPORT

A 24 year old gravida 3, para 1, abortus 1 female, first presented to our clinic at 23 weeks of gestation for routine control. She aborted a 6 weeks gestation 3 years ago and gave birth to a 2000 gr healthy male at the same year. The ultrasound scan at the first visit revealed a severe midline defect of the fetal abdominal wall and a large extra-abdominal mass containing bowel and liver (Figure 1). Severe scoliosis, clubfoot, absent urinary bladder and oligohydramnios were the other findings. All of these findings were suggestive of a body stalk anomaly. Termination was offered. The family felt they could not terminate the pregnancy and choose to continue. At 38 weeks of gestation patient admitted to our clinic with spontaneous active labor. With the indication of transverse situs, cesarean section was performed and a boy weighing 2000 g was born with an Apgar score of 2/2. Cardiopulmonary resuscitation was not successful and he died 30 minutes after birth. Macroscopically, abdominal organs were in a sac covered by the amnion and were attached directly to the placenta. Umbilical cord was 8 cm in length. Other clinical findings were scoliosis, clubfoot, absence of left lower extremity and both kidneys. The family did not permit for the autopsy. X-ray graphies and morphologic appearance of the fetus are shown in Figure 2 and 3.

DISCUSSION

LBWC is a rare polymalformation presenting as a combination of at least two of the following three features: exencephaly; and facial clefts, thoraco- and/or abdominoschisis (midline defect); and limb defect.[1,3] The wide phenotypic spectrum of defects has given the disorder many names including amniotic band syndrome, body stalk anomaly and limb body wall complex. There is no general consensus on the most appropriate name, or on the most likely etiology.

The pathogenesis of body stalk anomaly is uncertain. There are several mechanisms explaining the development of body stalk anomaly. One of them is early amnion rupture before obliteration of the coelomic cavity, with the passage of the lower half of the fetal body into the coelomic cavity through the defect in the amniotic sac.[6] The other suggested mechanism is the early generalized compromise of embryonic blood flow during the first 4-6 weeks of gestation, leading to failure of closure of the ventral wall and persistence of the extra-embryonic coelomic cavity.[1, 2] The most commonly accepted hypothesis is abnormal embryonic folding. This hypothesis was initially suggested by Streeter in 1930.[7] It's supported by other authors also.[4, 8-11] During the 5th week of gestation, the trilaminar embryo, which is connected to the placenta by a body stalk, is transformed into a cylindrical fetus by folding in cephalic, lateral and caudal axes. Body stalk anomaly is due to faulty folding in all three axes, with persistence of the extra-embryonic celomic cavity. The various malformations associated with body stalk anomaly depend on the degree of aberrant development of each of the four folds.

Figure 1. Sonographic appearance of abdominal wall defect.

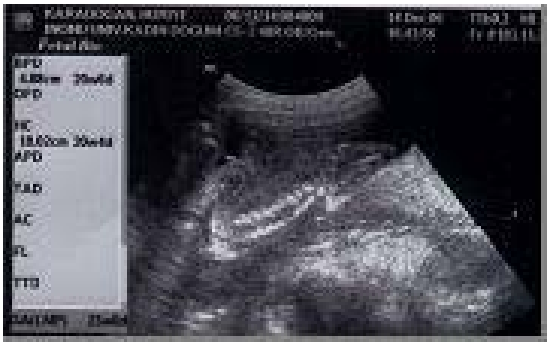
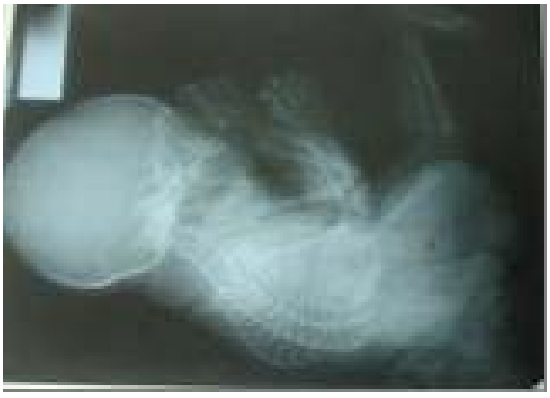


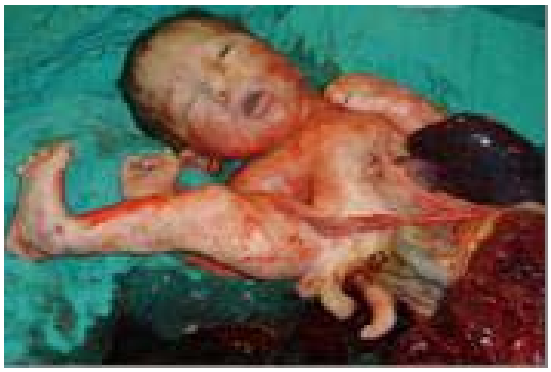
Figure 2. Scoliosis and absent left lower extremity



Body stalk anomaly is usually diagnosed during the second trimester of pregnancy by ultrasound carried out for gestational age dating or for evaluation of an elevated maternal serum α-fetoprotein. Body stalk anomaly is diagnosed sonographically by showing kyphoscoliosis, short umbilical cord and a large abdominal wall defect with evisceration of both the

liver and bowel. Limb deficiency and/or exencephaly may or may not accompany. Although difficult due to the normal physiological mid-gut herniation of bowel, it can also be diagnosed during the first trimester.[4,9] Smrcek et al [11] in a retrospective study determined the sonographic findings of body stalk anomaly in 11 cases between 1994 and 2001. None of the 11 fetuses showed craniofacial defect. Six fetuses presented with abdominoshisis and five with thoracoabdominoshisis with eventration of different thoracic and /or abdominal organs. Neural tube defect (spina bifida) was diagnosed in three cases. Kyphoscoliosis was diagnosed in 9 fetuses. Limb abnormalities including arm amelia, split foot, clubfoot and malposition of the lower limb were present in 6 fetuses. All placental examinations showed evidence of persistence of the extraembryonic celomic cavity. The umbilical cord was malformed and short in all cases and in five fetuses color Doppler examination revealed a single umbilical artery. Sonography done in the first trimester may reveal embryo abdomen finding similar to omphalocele but the lower embryonic body is found to be in the celomic cavity in the body stalk anomaly.[12] Furthermore the nuchal translucency thickness was above the 95th percentile of the normal range for crown-rump length in 71.4% of the cases.[4]

Figure 3. Morphologic appearance of the fetus



The cause of body stalk anomaly is not found. Fetal karyotyping by amniocentesis and/or chorion villus sampling (CVS) was found to be normal.[4, 11] In one case CVS showed a mosaic trisomy 2 in the placenta.[11] In another case molecular study of polymorphic markers for chromosome 16 on fetal tissue revealed maternal meiotic error in the placenta and uniparental disomy for chromosome 16 in the fetus.[13] It's stated that maternal uniparental disomy for chromosome 16 and mosaic trisomy 2 in the placenta, by causing placental insufficiency can be responsible in the etiology of body stalk anomaly.

These infants usually die soon after birth because of severe pulmonary hypoplasia and most obstetricians consider this anomaly invariably fatal, even today.[14] However a few babies with body stalk anomaly have been saved.[15, 16] With the remarkable progress in intensive care strategies for fetuses and neonates, the number of survivors may increase in the future.

CONCLUSION

Body stalk anomaly is a rare type of anterior wall defect and can be confused with omphalocele and gastroschisis sonographically. Although few babies with body stalk anomaly have been saved, it's accepted as a fatal anomaly. It's important to distinguish body stalk anomaly from other anterior wall defects to determine the management options.

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