

A Fetal Autopsy Case of Body Stalk Anomaly: Case Report

Body Stalk Anomalili Bir Fetal Otopsi Olgusu

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ABSTRACT Body Stalk Anomaly is a rare, fatal anomaly characterized by an abdominal wall defect, scoliosis and a short umbilical cord. In this report a case of body stalk anomaly diagnosed at autopsy is presented. A 26-year-old pregnant woman presented to this hospital for routine antenatal ultrasonography scanning. Ultrasound scans revealed a fetus with the gestation age 20 weeks that has multiple congenital defects and therapeutic abortus was done. Fetal autopsy revealed a large omphalocele sac containing most of the abdominal organs, conspicuous scoliosis, clubfoot anomaly and short umbilical cord. Since body stalk anomaly is incompatible with life, pregnancy termination should be considered. It is important to differentiate it from other anterior wall defect and early prenatal diagnosis for evaluating the management options.

Key Words: Hernia, umbilical; scoliosis; clubfoot

ÖZET "Body stalk" anomalisi, karın ön duvar defekti, skolyoz, kısa göbek kordonu ile karakterize nadir görülen fatal bir anomalidir. Burada tanısı otopsi ile konulmuş body stalk anomalili olgu sunuldu. Yirmi altı yaşında gebe, rutin antenatal ultrasonografi taraması için hastanemize başvurdu. Ultrason ile incelemede multipl konjenital anomalilere sahip 20 haftalık fetus gözlendi ve terapötik tahliye yapıldı. Fetal otopside başlıca karın iç organlarını içine alan büyük omfalosel kesesi, belirgin skolyoz, yumru ayak anomalisi, kısa göbek kordonu gözlendi. Body stalk anomalisi hayatla bağdaşmadığından gebeliğin sonlandırılması düşünülmelidir. Diğer anterior duvar defektlerinden ayırıcı tanısı ve erken prenatal tanısı tedavi planının belirlenmesi açısından önemlidir.

Anahtar Kelimeler: Fıtık, umbilikal; skolyoz; yumru ayak

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Body stalk anomaly refers to a variable group of congenital defects having in common abdomino- or thoraco-schisis and limb defects. The synonyms of body stalk anomaly are limb-body wall complex or aplasia of the cord. This congenital anomaly has no sex or familial predilection or known recurrence risk. Karyotype study has been reported to be normal in all cases of body stalk anomaly. The incidence of body stalk anomaly is estimated to be 0,7:10000 births.¹ This anomaly is invariably fatal.^{1,2} In this case report postmortem findings of fetus with typical body stalk anomaly is presented with reference to studies in literature.

CASE REPORT

A 26-year-old woman applied for routine pregnancy examination to our hospital. She was G1P0A0, at the time of scanning. She gave a history of a non-consanguineous marriage, past medical history was unremarkable and the patient denied the use of cigarette, alcohol or drugs. Ultrasound scans revealed a fetus with derived gestational age as 20 weeks 4 days and biparietal diameter as 20 weeks 2 days. Multiple anomalies were determined in ultrasound examination. The scan revealed a large ill-defined abdominal wall defect, evisceration of the abdominal organs into an omphalocele sac (Figure 1). There was evidence of scoliosis and hypoplasia of thorax. Ultrasonographic views of dilatation of ureter and hydronephrosis were shown in Figure 2. The patient elected to terminate the pregnancy. Postmortem physical examination showed a fetus with large (4.5 x 4 cm)

omphalocele sac in the abdominal area. Placenta was adhered to the omphalocele sac in one region. Umbilical cord was 7 cm in length (Figure 3). Umbilical cord section showed an artery and a vein. There was evidence of scoliosis with conspicuous angulation and convexity to the left (Figure 4). There were 2 skin tags in the genital area and there was no discernible external genitalia and urethra. Anal atresia was present (Figure 5). Pes equinovarus was found in right extremity. Club foot anomaly was found in left extremity. When omphalocele sac was dissected serous fluid was evacuated. Inside the sac; liver, stomach, small and large intestine, pancreas and spleen were detected. There was evidence of bilateral abdominal testis. The diaphragm was absent. In retroperitoneal examination the left kidney was larger than normal and showed pelvicaliceal dilatation. The left ureter was conspicuously dilated. Both of the ureters



FIGURE 1: View of omphalocele.



FIGURE 2: View of dilatation of ureter.



FIGURE 3: Anterior view of the fetus. The figure shows omphalocele sac, short umbilical cord, clubfoot, pes equinovarus deformities in low extremities. It also shows placenta adhered to the omphalocele sac.



FIGURE 4: Posterior view of the fetus with body stalk anomaly shows marked scoliosis (convexity to the left).



FIGURE 5: The figure shows indeterminate external genitalia with bilateral streak gonads reminiscent of skin tags (arrow) and anal atresia.

were seen opened to the bladder which was found on the omphalocele sac wall. When thorax cavity was opened it was seen to be hypoplastic. The left lung was shown to be pushed behind the back wall of the thorax and it also showed hypoplasia. There was no cardiac malformation. When skull was opened there was a brain with two hemispheres. Corpus callosum was absent. Lateral ventricles were dilated. According to body sizes the maturation was compatible with 18th weeks of gestation. In the light of all the findings the diagnosis was body stalk anomaly. Informed consent was obtained from the patient.

DISCUSSION

Body stalk anomaly is a rare fetal polymalformation of uncertain etiology. The wide phenotypic spectrum of defects has given the disorder many names including limb body wall complex, body stalk anomaly, body wall complex, amniotic band syndrome, cyllodomas, congenital absence or aplasia of the umbilical cord. There is no general consensus on the most appropriate name.

Van Allen et al described this anomaly based on the existence of three criteria; a) exencephaly or encephalocele with facial clefts b) thoraco and/or abdominoschisis c) limb defects. The diagnosis was based on the presence of two out of three above abnormalities.² Our case showed abdominal wall and limb defects.

The pathogenesis of the body stalk anomaly is unclear, although there are several mechanisms explaining the development of this anomaly. First causative theory is early amnion rupture before obliteration of the coelomic cavity, primary rupture of amnion leading to formation of amniotic bands.³ Second suggested theory is vascular disruption during the first 4-6 weeks of gestation.² Another theory is germ disc defect with early embryonic maldevelopment.⁴ The last theory is the most commonly accepted.^{5,6}

Russo et al have proposed that there are two distinguishable phenotypes: the “placento-cranial” and “placento-abdominal” adhesion phenotypes. The first phenotype shows craniofacial defects and/or adhesion, while the second is without craniofacial defect and present urogenital anomalies, anal atresia, abdominal placental attachment.⁷ The pattern of our case resembled the placento-abdominal adhesion phenotype.

Viscarello et al and Negishi et al reported that cases of body stalk anomaly were related to cocaine abuse.^{1,7} In a study of a series of fetuses with body stalk anomaly by Luehr et al; in 50%, 50% and 30% of women a history of cigarette, alcohol and marijuana use, respectively, was noted.⁸ Our patient denied the use of cigarettes, alcohol and cocaine.

Although according to some reports body stalk anomaly may show recurrence, the risk of recurrence of body stalk anomaly is negligible.⁸ In our patient second baby was healthy.

Van Allen in a study of a series of fetuses with body stalk anomaly found that the major structural defects included limb defects (95%); marked scoliosis (77%), internal organ malformations (95%) and craniofacial defects (56%). The internal organ malformation present in 95% of cases was cardiac defects, absent diaphragm, bowel atresia, renal agenesis and renal displasia.² Smrcek et al in a study of a series of 11 cases with body stalk anomaly found that the defects included kyphoscoliosis in 9 fetuses, abdominoschisis in 6 fetuses, thoracoabdominoschisis in 5 fetuses and neural tube defects in 3 fetuses. Limb abnormalities included arm amelia, split foot, club foot and malposition of the lower

limb. There was no craniofacial defect in their series.⁶ The umbilical cord was malformed and short in all cases and five fetuses showed single umbilical artery. In another large series Van Allen et al found body defect in 96% of cases. The involvement of both abdomen and thorax was a more common feature as compared involvement of either abdomen or thorax solely. Our case had a midline abdominal wall defect, marked scoliosis and a short umbilical cord. We observed complete absence of external genitalia and anal atresia in our case.

The prognosis of body stalk anomaly is very poor compared to isolated omphalocele or gastroschisis. Early diagnosis of body stalk anomaly play an important role since prognosis is very poor, termination must be applied. Liveborn infant die shortly afterward. Ustun et al reported a fetus in 38 weeks of gestation with body stalk anomaly who died 30 minutes after cesarean section.⁹ Only a few infant with body stalk anomaly have been saved and the condition is still generally considered fatal because of the severe pulmonary hypoplasia.¹⁰

Body stalk anomaly is diagnosed by two-dimensional and three-dimensional ultrasonography, mostly in the second trimester of pregnancy during gestational age dating. It can also be diagnosed during first trimester⁵. In the literature there is a report of a case that is diagnosed during 9 weeks of gestation.¹¹ There are cases which have been diagnosed autopsy.¹² In our case; multiple anomalies were detected during ultrasonography scanning before therapeutic abortus. The final diagnosis as body stalk anomaly was based on autopsy findings.

This case report provides a shortly review of body stalk anomaly, including the diagnosis, pathogenesis, prognosis and associated malformations. Omphalocele is a prominent sonographic marker for this anomaly. Prenatal detection of abdominal wall defect associated with severe scoliosis, short umbilical cord should alert the clinician and pathologist to the possibility of body stalk anomaly.

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