Body stalk anomaly – case report and review

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Abstract

Body stalk anomaly is a term used to describe a pattern of severe defects that are incompatible with life. This condition should be suspected when encountering a large abdominal defect, along with axial skeletal abnormalities such as kyphosis or scoliosis and a short or absent umbilical cord. Body stalk anomaly is considered a fatal anomaly, thus being crucial to distinguish it from other anterior wall defects when evaluating the management options. We report the case of a patient diagnosed at 15 weeks of gestation with a complex of severe fetal anomalies that were confirmed after the termination of pregnancy as body stalk anomaly, and we also discuss data published in literature regarding this topic.

Keywords: *body stalk anomaly, ultrasound, lethal*

Rezumat

Body stalk anomaly reprezintă un complex de anomalii fetale care sunt considerate incompatibile cu viața. Această anomalie ar trebui suspectată în cazurile în care se identifică un defect abdominal major împreună cu anomalii scheletale precum cifoză sau scolioză și cordon ombilical scurt sau absent. Având în vedere prognosticul sever, este foarte important diagnosticul diferențial cu alte defecte de perete abdominal anterior, pentru a putea oferi pacientei o consiliere adecvată. În lucrarea de față, prezentăm un caz diagnosticat cu această anomalie la vârsta gestațională de 15 săptămâni și, de asemenea, trecem în revistă datele publicate în literatură despre acest complex malformativ.

Cuvinte-cheie: *body stalk anomaly, ecografie, letal*

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Body stalk anomaly – prezentare de caz și discuție

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Introduction

Body stalk anomaly is a rare congenital defect with an incidence rate ranging from 1 in 14,000 to 1 in 31,000 pregnancies, according to large epidemiological studies. However, a recent study, evaluating 106,727 fetuses between 10 and 14 weeks of gestational age, reported a rate of approximately 1 in 7500 pregnancies, considering the increased incidence of miscarriages associated with this condition[1,2].

Body stalk anomaly refers to a group of severely disfiguring abdominal wall defects where the abdominal organs protrude outside the abdominal cavity contained within a sac of amnioperitoneum, often with the absence or with a very small umbilical cord.

Potential causes may include early amnion rupture due to direct mechanical pressure and amniotic bands, vascular disruption of the early embryo, or abnormalities in the germinal disk[3,4].

Case report

We present the case of a gravida 2, para 2 patient with no relevant medical history who was referred to our maternal-fetal department for an ultrasound scan at 15 weeks of gestation. We did not identify any teratogenic risk factors from her clinical history, and her first pregnancy and delivery were uneventful. She hadn't done the first-trimester anomaly combined screening, nor the non-invasive prenatal test (NIPT).

The ultrasound scan revealed complex fetal malformations involving a large anterior abdominal wall defect with herniation of viscera – i.e., the liver and bowel loops through the defect which appeared to be attached to the placenta. The heart had a normal position within the thoracic cavity. The lumbosacral spine also appeared abnormal, with the visualization of normal vertebrae only in the cervicothoracic region. Only one lower limb was observed and, on the other side, a rudimentary structure of the other limb. The fetal torso appeared to be immobile, attached to the placenta (Figure 1).

The fetal skull, face and upper limbs appeared to be normal.

Having counseled the patient that this malformation is incompatible with life, she chose to terminate the pregnancy, and the procedure was performed without complications. An examination of the fetus revealed eviscerated abdominal organs adherent to bands of amniotic tissue and confirmed the ultrasound findings presented before (Figure 2).

Discussion

The most accepted theory regarding the development of body stalk anomaly is that it originates from a defect in the germinal disk[5].

During the fifth week of gestation, the flat trilaminar embryo undergoes transformation into a cylindrical fetus through the formation of four parallel, contiguous body folds: cephalic, caudal, and both lateral folds. Malformation of any of the four folds leads to a specific combination of anomalies. Anomalies in

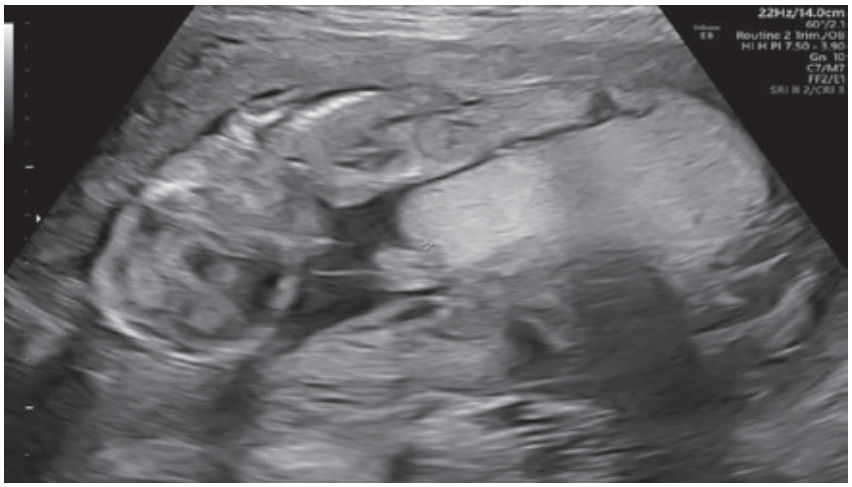

Figure 1. *Abnormal position of the fetus which appeared immobile and attached to the placenta, having a very short umbilical cord*

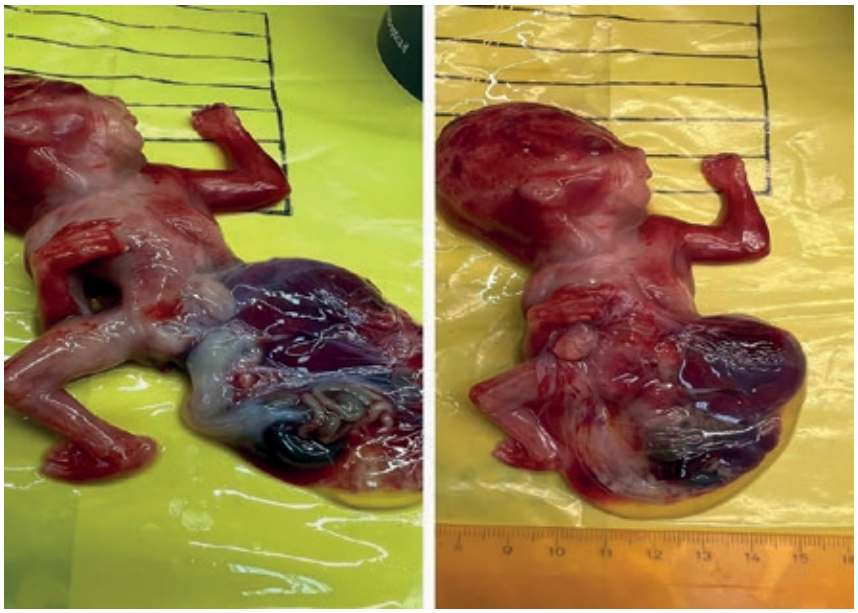

Figure 2. *Fetus with large abdominal wall defect, with herniation of the viscera. A very short umbilical cord was observed and also only one lower limb. Both upper limbs and face were normal*

cephalic folding can lead to a presentation resembling the pentalogy of Cantrell, defects in lateral folding may result in omphalocele, and aberrant caudal folding may cause various abnormalities seen in cloacal exstrophy[6-8].

The common ultrasound findings include a significant defect in the abdominal wall, severe kyphoscoliosis, limb abnormalities, neural tube defects, and a malformed, short umbilical cord.

Usually, the fetuses do not display craniofacial defects. These elements are all consistent to the ones observed in our patient. The examination of the placentae reveals signs of persistent extra-embryonic celomic cavity.

Additionally, NT measurements are usually abnormal. The AFP level in amniotic fluid tends to be increased in almost all cases[9,10].

The fetus is restricted in movement and appears almost fused with the placenta, similar to our case. Limb amputations and encephalocele could result from the entrapment of fetal limbs and the skull, respectively, within the celomic cavity[11].

The list of possible conditions to consider alongside body stalk anomaly includes other fetal abdominal wall defects such as:

- Amniotic band syndrome (random defects; constriction rings; amputations; bands).
- Gastroschisis (small defects; without membrane; normal cord insertion; bowel complications)[12].
- Omphalocele (variable-sized defects; abnormal cord insertion; membrane-covered liver/bowel; ascites).
- Pentalogy of Cantrell (sternal, pericardial, diaphragmatic defects; *ectopia cordis*; large omphalocele)[12].
- Beckwith-Wiedemann syndrome (organomegaly; polyhydramnios; macroglossia; large omphalocele).
- Bladder exstrophy (absence of fluid-filled bladder; splayed/absent pubic bone; soft tissue mass seen at the lower abdomen).
- OEIS complex (large omphalocele, exstrophy, imperforate anus, spinal anomalies complex)[13].

Typically, fetuses with body stalk anomaly have a normal karyotype, therefore routine karyotyping is not necessary in these cases. There is only one reported case in the literature of placental trisomy 16, maternal uniparental disomy 16 and body stalk anomaly, suggesting placental insufficiency or imprinting effects as the cause of this anomaly[14].

Conclusions

Body stalk anomaly is a congenital pathological condition characterized by uncertain etiopathogenesis, pathophysiology and incidence rate, but with a very poor prognosis.

We consider reporting another case with this severe malformation important, as it marks the role that early recognition of ultrasound signs has in establishing the correct diagnosis, along with the appropriate counseling of the couple and prompt management by physicians. ■

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