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BODY STALK ANOMALY IN A 14-WEEK PREGNANCY: A CASE REPORT

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ABSTRACT Body stalk anomaly is an uncommon and dangerous malformation for which there is no recognized cause or precise etiology. This is a case of a 29-year-old patient whose ultrasound at 12 weeks indicated megacystis and fetal ectopia cordis. A second sonography was performed on the patient to confirm the diagnosis. The results indicated a short umbilical cord, an angulation deformity of the spine, the absence of the mid and distal segments of one upper limb, ectopia cordis, a significant abdominal wall abnormality consistent with omphalocele and the presence of a cloaca. The ultrasonographic results supported body stalk anomaly. Her dual marker screen was positive for Trisomy 18/13. Because of serious deformities that were conflicting with life, the patient was given the option of terminating the pregnancy. The suspected anomaly was confirmed on pathologic examination.

KEYWORDS : abdominal wall defect, body stalk anomaly, limb-body wall complex

Background

The most uncommon and dangerous type of abdominal wall abnormality is body stalk anomaly. The peritoneal cavity is accessible to extraembryonic coelom in this condition, which results in the disembowelment of abdominal organs and, in extreme cases, thoracic organs since the abdominal wall fails to mature. This malformation is accompanied by undeveloped umbilical cord which may be short or absent1. Severe kyphoscoliosis is also associated with it1. It may also occur concomitantly with neural tube defects, genitourinary malformations, chest wall abnormalities, craniofacial distortions and intestinal atresia2,3. These variety of presentations reported in cases worldwide have created confusion between terms like amniotic band syndrome, short cord syndrome and limb-body wall complex4. The estimated incidence is between 1/14,000 to 1/42,000 pregnancies5. However, the newest research conducted by Daskalakis et al. reported an incidence of 1/7,500 pregnancies after analyzing 1,06,727 fetuses between 10 to 14 weeks1. Due to the disparity in occurrence rates, it is possible that this deformity is to blame for a significant percentage of first trimester spontaneous miscarriages.

The actual mechanism of possible trigger of this anomaly is unknown. Different theories have been put forward. The impacted fetuses' karyotypes have been proven to be normal6. Chromosomal aberrations have been recorded in only 2 cases till date i.e., with uniparental disomy of chromosome 16 and with trisomy of chromosome 26. Environmental and hereditary factors may influence the disease process, but it is yet to be clearly understood[7].

The differential diagnosis of body stalk anomaly are other abdominal wall deformities like gastroschisis (minor defects; devoid of membrane; usual cord insertion; bowel complications), omphalocele (varying sized defects; atypical cord insertion; membrane protected bowel/liver; ascites), amniotic band syndrome (incidental defects; constrictions; bands) , Pentalogy of Cantrell (sternal; pericardial; diaphragmatic deficiencies; ectopia cordis; sizeable omphalocele), Beckwith-Wiedemann syndrome (organomegaly; polyhydramnios; macroglossia;signifi cant omphalocele), bladder exstrophy (missing fluid-filled bladder; distorted/nonexistent pubic bone; soft tissue mass in lower abdomen) or OEIS complex (significant omphalocele; exstrophy; imperforate anus; spinal anomalies complex)[8,9].

Case description

We report a case of a 29-year-old Indian woman who attended our OPD for her first antenatal checkup at 12 weeks of gestation. This was her second gestation, and she had a history of caesarean section 7 years back with no antenatal complications and delivered a healthy live issue. Her initial prenatal lab reports suggested a high blood sugar level and HbA1c level of 10.2 percent. Other lab reports were within acceptable range.

At 12 weeks of pregnancy, she had her first sonographic assessment which reported a normal fetal crown-rump length of 4.78 cm with ectopia cordis and megacystis. A second sonography was performed on the patient to confirm the diagnosis. The resulting pictures indicated that the inferior body was located in the coelomic space, a short umbilical cord, single umbilical artery, angulation deformity of the spine, absent mid and distal segment of one of the upper limbs, ectopia cordis, a severe abdominal wall defect consistent with an omphalocele and a large cystic lesion of size 2.4 × 1.8 cm in fetal lower abdomen suggestive of cloaca. The amniotic fluid level was normal. Nasal bone was present, nuchal translucency was 1mm but ductus venosus was not delineated. The ultrasonographic results supported body stalk anomaly (figures 1 and 2).

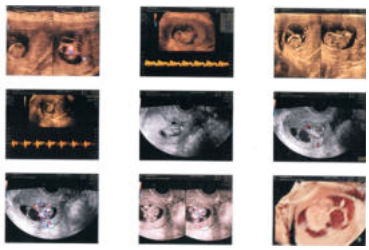

Figure 1: Ultrasonographic image suggesting the location of inferior body into coelomic space.

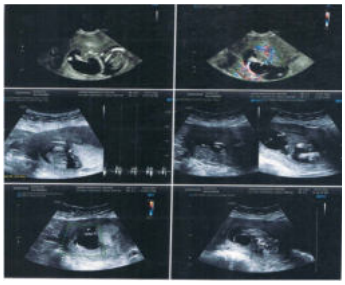

Figure 2: Image showing angulation deformity of the spine, absent mid and distal segment of one of the upper limbs, ectopia cordis, a severe abdominal wall defect consistent with an omphalocele and a large cystic lesion in fetal lower abdomen.

Dual marker screen was positive for Trisomy 18/13 with risk probability of 1:24.

The patient was given the option of voluntarily terminating the pregnancy at 14 weeks because the abnormality was thought to be

incompatible with life. The fetus was sent for pathologic examination after medical termination. The findings of examination were as follows: anterior abdominal wall defect with organs (liver, spleen and intestines) protruding out as masses of tissues clumped together. Anus was imperforate. The thoracic cavity was empty. Heart, lungs and diaphragm were absent. Kidneys and supra-renal tissue were ill-defined. A cyst was present in the sacral region. Kyphoscoliosis was present and the left limb was dislocated backwards. (Figure 3) The couple refused to get a cytogenetic study done.

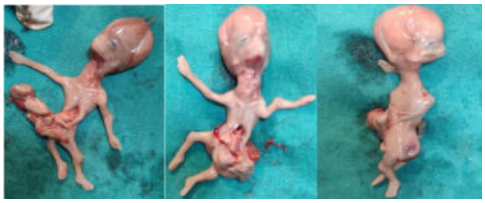

Figure 3: Anterior abdominal wall defect with organs protruding out as masses of tissues clumped together, left upper limb dislocated backwards & Kyphoscoliosis of spine.

Discussion

A body stalk anomaly must be contemplated when there is a significant abdominal wall defect, axial skeleton abnormalities including kyphosis or scoliosis, and a short or nonexistent umbilical cord as described in the above case10. It has been established that the majority of reported cases are not compatible with life.

An ultrasound can find these anomalies late in the first trimester. In our case, the diagnosis was made at 12 weeks and confirmed on a later ultrasound. The sonographic results matched those that were stated in the literature. In certain cases, this anomaly has been reported in 50% of fetuses of women who smoked cigarettes or drank alcohol and in 30% of marijuana or cocaine abusers11. But in our situation, we found no substantial risk factor linked to exposure to teratogens in the environment. Measuring levels of maternal serum alpha-fetoprotein mainly in the second trimester might detect 100% of cases as reported by some authors[12].

Three key explanations can account for the main phenotypic characteristics of the body stalk abnormality13. The best acknowledged concept is premature rupture of the amnion prior to obliteration of coelom13. A second idea contends that aberrant trilaminar disc folding in the cephalic, caudal, and lateral orientations causes the coelomic cavity to persist as proposed by Steetar et al14. Lastly, the third theory proposed by Van Allen et al. suggests that a vascular compromise in early weeks of pregnancy could result in early amnion rupture[13,15,16].

There is no recognized cause or precise etiology for this anomaly. It is believed that the germinal disc defect results in unsuccessful body folding along all three axes (cephalic, caudal and lateral)17. Typical body folding leads to the division of the intraembryonic and extraembryonic coeloms, the production of the body stalk, and the normal formation of the umbilical cord. A thoracic wall and epigastrium deformity brought on by abnormal cephalic folding results in ectopia cordis. When the lateral folds are abnormal, the abdominal contents protrude into the amnio peritoneal sac, which unites peripherally to the chorionic plate without an umbilical cord or with a short cord. Severe scoliosis results from asymmetrical development of spine and thoracic cavity.

Literature describes two main phenotypes resulting from distinct pathogenic ways:

- The placental-cranial form involving craniofacial defects (encephalocele/exencephaly with facial clefts) and amniotic bands between cranial defects and placenta resulting from early vascular disruption[18,19].

- The placental-abdominal form involving urogenital anomalies, anal atresia, lumbosacral meningocele, short cord, persistence of extraembryonic coelom and intact amnion resulting from anomalous embryonic development[18,19].

In our case the predominant phenotype is placental-abdominal with both abnormal cephalic and lateral folding resulting in ectopia cordis, persistence of extra-embryonic coelom and a short cord but no craniofacial defects. So, the pathogenetic mechanism here is embryonic maldevelopment.

Clinical Significance

The patient's dual marker screen was positive for Trisomy 18/13 with a risk probability of 1:24. Unfortunately, the couple refused to get a cytogenetic study done which could have been invaluable evidence in establishing the relation between the anomaly and its genetics. The sonographic results matched those that were reported in the literature. The early diagnosis of body stalk anomalies with ultrasound towards ending of the first trimester is crucial for patient management because they are incompatible with life. Early detection gives parents the knowledge they need about the prognosis of this abnormality.

Acknowledgement

Nil

Statement of ethics

The study abided with the principles outlined in the Helsinki Declaration. The patient provided written informed consent in order for this case report to be published.

Conflict of interests

The authors affirm that they have no competing interests with regard to the publication of this research.

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