Case Report

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Body Stalk Deformity During First Trimester: A Rare Reporting

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Abstract

Body stalk anomaly is a rare congenital anomaly, its etiology, pathophysiology and predisposing factors are not yet known. It is a combination of multiple development abnormalities including body wall defect and can be associated with spine, limb defects. It can be associated with multiple syndromes like amniotic band syndrome, Limb body wall complex. We represent a case of body stalk deformity at a gestation of 13 weeks which was proven post termination of pregnancy. Body stalk anomaly is a type of Limb Body Wall Complex and should be differentiated from other body wall defect as its prognosis is poor as compared to individual body wall defect.

Keywords: Body stalk anomaly; Limb defects; Body wall defect.

INTRODUCTION

Body stalk anomaly is a rare fetal sporadic polymalformativeanomaly associated with anterior abdominal wall defects. Body stalk anomaly should be differentiated with other abdominal wall defects such as omphalocele, gastrochiasis, ectopiacordis, pentalogy of Cantrell and OEIS complex.[1,2]

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A study by Daskalakis *et al*, in gestation of 10-14 week 106,727 fetuses analyzed, and prevalence of 1/7,500 pregnancies was found. As body stalk anomaly is a rare syndrome with poor prognosis the reported case is less as this can lead to spontaneous abortion in first trimester before diagnosing. Few studies reported.[12] cases per 10,000 births or one in 14,000–22,000 pregnancies. The live births are very less about 11/428,599 births or about 0.2-0.3 per 10,000 births. Body stalk anomaly can be associated with placental trisomy 16 or maternal uniparental disomy 16 however usually it is not associated with chromosomal abnormalities and karyotyping is normal. There is no association with the fetal gender of age of the parents. Also the chances of recurrence in subsequent pregnancies are very low.[1,3,7]

Typical features of body stalk anomaly involves abdominal wall defect with herniation of abdominal viscera, spinal abnormalities like kyphoscoliosis, short or absent umbilical cord (UC), increased nuchal thickness. It can be associated with amniotic band syndrome as seen in our case. Complex body

wall anomalies include body stalk anomaly and limb body wall complex. Limb body wall complex involve body wall defect as well as limb defect and can be associated with craniofacial abnormalities like cleft palate *etc*.[4-6] There is abdominal wall defect in the body stalk anomaly, wall is not formed so the peritoneal cavity is open with herniation of liver and intestine into the extraembryonic coelom.[3,8]

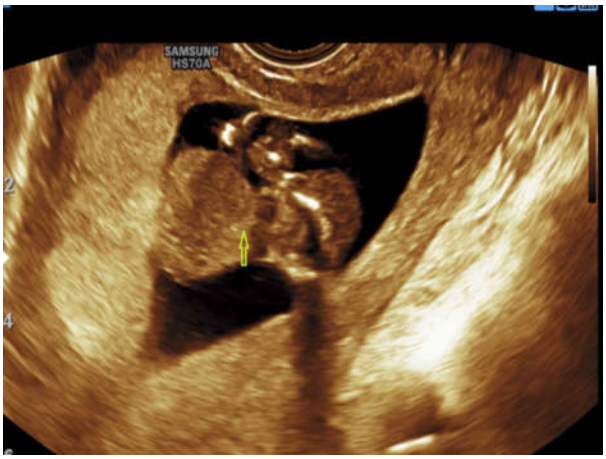
CASE REPORT

A 29-year-old female G4 P2 L1 was referred to our radiology department to undergo a nuchal translucency scan at 13 weeks'of gestation. She had two miscarriages in the past at around 5-6 weeks of gestation of unknown etiology. Patient karyotyping was done which was normal. However fetal karyotype was no evaluated as the patient was not willing. No further details were known. She reported no relevant medical history (Diabetes, Hypertension). Patient gave no history of drug abuse, chronic medication or infectious disease. There was no history of alcohol or tobacco use in any form of throughout the duration of pregnancy. She also had a normal blood profile.

A detailed transvaginal ultrasound scan revealed up fetus with the crown rump length of 53 mm consistent with the gestational age of 12 weeks. The scan revealed a large abdominal wall defect with abdominal content herniated into extra embryonic coelom. Body Stalk can involve Chest and Abdomen, however in our case the thoracic cavity appeared normal.

The Eviscerated organs formed a complex mass consisting of liver, stomach and bowel loops. There was evidence of Kypho-scoliosis of spine. Although four limbs were present, umbilical cord was short and single umbilical artery was seen. Fetal head circumference was normal. The abdominal diameter was disproportionally reduced. There was no anomaly seen in the fetal eye, palate, lips and neck. The placenta was anterior and separated from the fetus. Amnioticfluid was adequate. On color Doppler single umbilical artery was seen and bilateral uterine arteries shows normal flow with normal PI. This is a case of Body Stalk Anomaly type of Limb Body Wall Complex as we have eviscerated organs, Kypho-scoliosis of Spine and short umbilical cord with single umbilical artery. And there no craniofacial and limb abnormalities.

Thin liner amniotic band were seen. No evidence of ectopic coris was seen. The patient was informed of the poor prognosis of the anomaly and after seeking informed consent, pregnancy was terminated. An autopsy was not performed because of lack of consent. However, post termination, eviscerated abdominal organs, short umbilical cord was found and limbs were normal, no anal atresia was seen at the time of birth as detailed by the physician.



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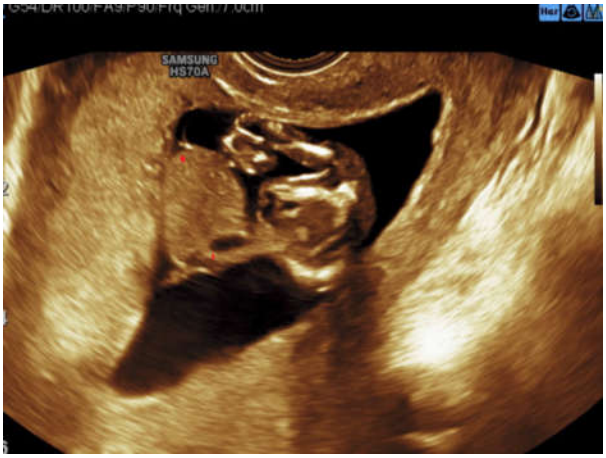

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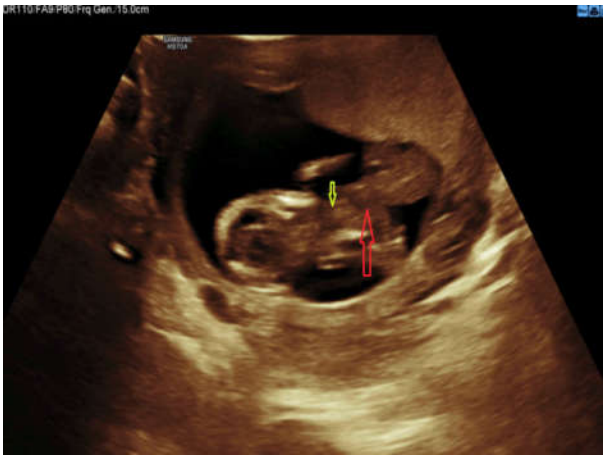

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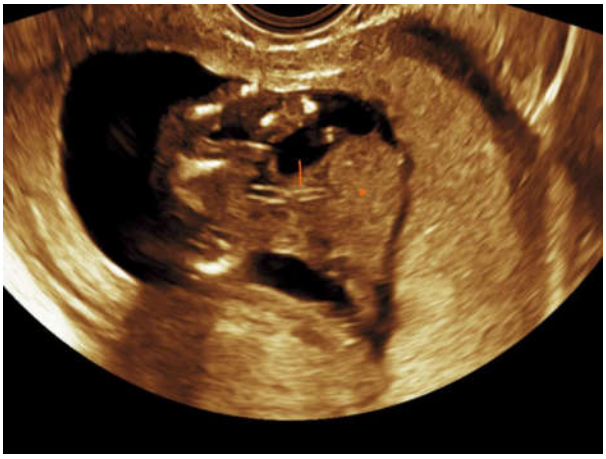

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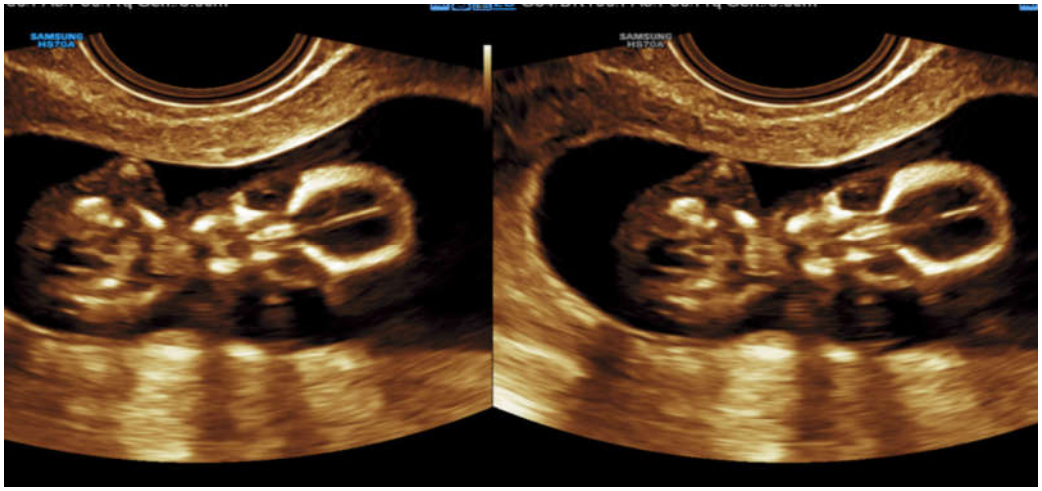

(E)

Transvaginal sonogram of a patient with 13 week gestation was done. (A) shows a defect in the anterior abdominal wall (arrow) with herniation of visceral organ into the amniotic cavity. (B) herniated stomach bubble (dot) and liver with intestine is seen (star) through the defect. (c) shows reduced dimension of the abdominal circumference (small green arrow) with herniated sac (red big arrow). (D) shows a small umbilical cord (linear red line) attached to herniated sac (dot). (e) shows normal appearance of face with no craniofacial defect.

DISCUSSION

Van Allen *et al*., in 1987 described the criteria for limb body wall complex (1) Exencephaly and/or encephalocele with facial clefts, (2)Thoraco and/or abdominoschisis, and (3) Limb defects. The other names associated with it are "Body stalk anomaly" "Congenital absence of umbilical cord" and "cyllosomus and Pleurosomus".[10,11]

Types of body stalk anomalies

According to the anatomy and embryology Martín-Alguacil and Avedillo classify body stalk anomaly as:

(a) BSA Type I: fetus with spinal and umbilical cord defects, thoracoabdominoschisis, anal atresia and/or other internal organ structural defects, and structural limb defects;

(b) BSA Type II: fetus with spinal and umbilical cord defects, thoracoabdominoschisis, anal atresia and/or other internal organ structural defects, and nonstructural limb defects;

(c) BSA Type III: fetus with spinal and umbilical cord defects, abdominoschisis, anal atresia and/or other internal organ structural defects, and structural limb defects; and

(d) BSA Type IV: fetus with spinal and umbilical cord defects, abdominoschisis, anal atresia and/or other internal organ structural defects, and nonstructural limb defects.[4,9]

Various theories are proposed for pathogenesis of body stalk anomaly early amnion rupture[13,] vascular disruption[12], and embryonic mal-development.[14]

Early amnion rupture is most acceptable theory by Torpin[13] proposed multiple amniotic bands due to rupture of amnion which hampers in the embryogenesis and fetal body parts seen outside the peritoneal cavity in extra embryomic coelom.[1,15] In our case amniotic bands was seen which support the above theory. Vascular disruption theory or endogeneous theory proposed by Van Allen *et al*. 1987, negative impact on normal embryonic blood

and hinders with normal morphogenesis. Cocaine and nicotine abuse can lead to body stalk anomaly supports this theory.[2,12] In our case, no risk factors or teratogenic factors could be identified. Streeter in 1930 suggested embryonic mal-development.[1,14,15] Body stalk anomaly occurs due to defect in folds at all the three axes. Pentalogy of Cantrell occurs due to cephalic fold defect, Omphalocele occurs due to lateral fold defect and cloacal exstrophy due to caudal folding defect.[1,2]

Two phenotypes of body stalk anomaly aredescribed according to placental attachment to body parts:[1,15]

(1) Placento-cranial attachment: Van Allen *et al*. described vascular disruption theory in which craniofacial defects and amniotic bands or adhesions. Upper limb defects with thoracoschisis or exencephaly/ encephalocele can be seen.

(2) Placento-abdominal adhesions due to intrinsic embryonic maldevelopment can lead to lower limb defects, internal organ defects like absent diaphragm, bowel atresia, renal dysplasias, urogenital anomalies, anal atresia, short umbilical cord, and persistent extraembryonic coelomic cavity.

The body wall complex can be diagnosed on ultrasound examination as early as first trimester. Also measuring maternal serum alpha feto protein level can lead to clue to the anomaly. As body stalk complex has poor prognosis it should be differentiated from other subtypes of anterior abdominal body wall defect which are treatable.[11]

Subtypes of abdominal wall defects are: Omphaloceleanterior abdominal wall defect is seen with herniation of visceral organs like liver, bowel loops, and urinary bladder which are covered by parietal peritoneum.If diagnosed in second and third trimester prognosis is better (66-93% success rate).[16]

Gastroschisis, is a right sided paraumbilical hernia of bowel loop into the amniotic cavity without a membrane and associated anomalies are uncommon

Pentalogy of Cantrell is a rare entity with supraumbilical defect, lower sternal defects, anterior diaphragmatic defects, defect in the pericardium, cardiac ectopia, cardiac anomalies like ventricular septal defect.[16,17]

CONCLUSION

Body stalk syndrome is a fatal congenital anomalies involving multiple defects which leads to poor prognosis of the disease. Once diagnosed and differentiated from other abdominal wall defects, it is advisable to terminate the pregnancy.

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