Article published online: 2021-07-30

Prenatal diagnosis of body stalk complex: A rare entity and review of literature

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Abstract

Body stalk anomalies are a group of massively disfiguring abdominal wall defects in which the abdominal organs lie outside of the abdominal cavity in a sac of amnioperitoneum with absence of or very small umbilical cord. Various hypotheses proposed to explain the pathogenesis of limb body wall complex include early amnion disruptions, embryonic dysplasia, and vascular disruption in early pregnancy. Body stalk anomaly is an accepted fatal anomaly and, hence, its early diagnosis aids in proper management of the patient.

Key words: Abdominal defects; body stalk complex; limb body wall complex; ultrasound

Introduction

Body stalk anomalies are rare anomalies with abdominal wall defects and associated limb and spine anomalies. They have a wide range of differential diagnoses such as omphalocele and gastrochisis.It is very important to differentiate body stalk complex from these entities as body stalk complex is invariably fatal whereas an isolated omphalocele carries good prognosis. We present a rare case with classic features of this anomaly.

A 20-year-old healthy primigravida was brought to the hospital at 14 weeks of gestation for routine antenatal evaluation. There was no significant family history or drug intake. Per abdominal examination revealed fundal height corresponding to 12 weeks size and fetal heart sounds were present with a heart rate of 130 beats per minute.

Access this article online	
Quick Response Code:	Website: www.ijri.org
	DOI: 10.4103/0971-3026.150162

Gray-scale ultrasound (USG) revealed single live fetus at 14 weeks of gestation with grossly abnormal morphology. The fetus had skeletal deformity and a large ventral abdominal wall defect.

Patient was further evaluated using color Doppler and three-dimensional USG. Biometry revealed a femur length of 15 mm corresponding to a gestational age of 14 weeks. Fetal head could not be visualized. Umbilical cord was very short causing burying of the fetus into the placenta [Figure 1A and B]. A large midline defect of anterior abdominal wall was noted and a membrane-covered abdominal mass containing liver and bowel loops was seen herniating through the abdominal wall defect suggesting a large omphalocele [Figure 1C]. However, other abdominal organs including stomach, spleen and urinary bladder were within the body [Figure 2A]. Heart was in normal location within the thoracic cavity. Skeletal deformities were bilateral talipes and severely kyphoscoliotic spine [Figure 2B]. Amniotic fluid volume was adequate.

Three-dimensional USG revealed grossly malformed fetus suggesting the diagnosis of a body stalk complex [Figure 2C].

Patient was counseled regarding the fatal condition of the fetus and she opted for medical termination of pregnancy.

Examination of the fetus revealed a large omphalocele with herniation of the small intestine and liver. A very short umbilical cord was observed which was attached to the omphalocele. The lower limbs were deformed with bilateral talipes [Figure 3A]. Herniated organs were covered by amniotic membrane [Figure 3B]. Autopsy of fetus revealed liver and bowel loops within the herniated sac [Figure 3C]. Fetus also had anencephaly with open spina bifida having a large defect [Figure 4A and B]. The karyotype was normal (46 XX).

Discussion

Body stalk anomaly is a rare malformation syndrome with a reported incidence of between 1 per 14,000 and 1 per 31,000 pregnancies in large epidemiologic studies.[1,2]

Various hypotheses have been proposed to explain the pathogenesis of body stalk complex. Vascular disruption theory, also known as the endogenous theory, includes events that negatively influence normal embryonic blood supply, thereby interrupting normal morphogenesis.[3] Vascular disruption theory is supported by the finding that cocaine abuse may increase the risk of body stalk anomaly.[4] Early amnion rupture theory proposes that the anomalies in body stalk complex occur secondary to multiple amniotic bands which interrupt embryogenesis and the fetus lies outside the amniotic cavity.[5] Streeter theory of embryonic dysgenesis is due to improper histogenesis resulting in disturbances in embryonic folding process which leads to the development of body stalk complex.

Body stalk deformity occurs from complete failure of body folding along all three axes (cephalic, caudal, and lateral).[6] After the folding of embryo, the intraembryonic coelom is separated from the extraembryonic coelom, and the umbilical cord subsequently forms.[7] If this folding does not occur, then the extraembryonic cavity is not obliterated and the body stalk is missing resulting in congenital absence of the umbilical cord with a wide-based insertion of the large fetal amnioperitoneal sac onto the placental chorionic plate.[8]

Various malformations of body stalk anomaly depend on the degree of aberrant development of each of the folds.

The internal malformations of the urinary and genital systems are usually associated with body stalk complex. They suggest abnormal mesodermal development. Abnormality of the placodes may lead to maldevelopment of these systems.[9]

Body stalk complex presents as a combination of at least two of the following features: exencephaly, facial clefts, abdominoschisis, and limb defects.[3,10] Lower limb associated anomalies include club foot, polydactyly, syndactyly, oligodactyly, brachydactyly, and amelia.

Two main phenotypes have been described in literature as follows:

- Fetuses with craniofacial defects: They show two specific characteristics: (a) encephalocele or exencephaly always associated with facial clefts and (b) amniotic bands and/or broad amniotic adhesion between the cranial defect and placenta
- Fetuses without craniofacial defects: They often present with urogenital anomalies, anal atresia, lumbosacral

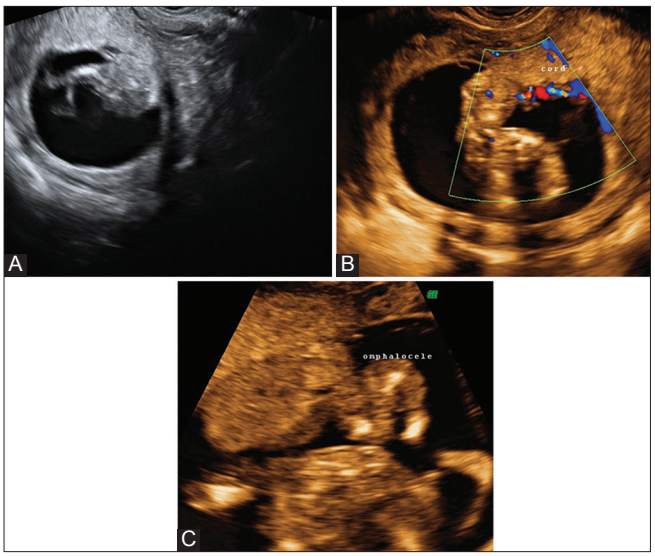

Figure 1 (A-C): (A) Gray-scale image reveals short umbilical cord with the fetus fixed to the placenta. (B) Three-dimensional ultrasound image with short umbilical cord resulting in fetus lying in close proximity to the placenta. (C) Three-dimensional ultrasound image with omphalocele

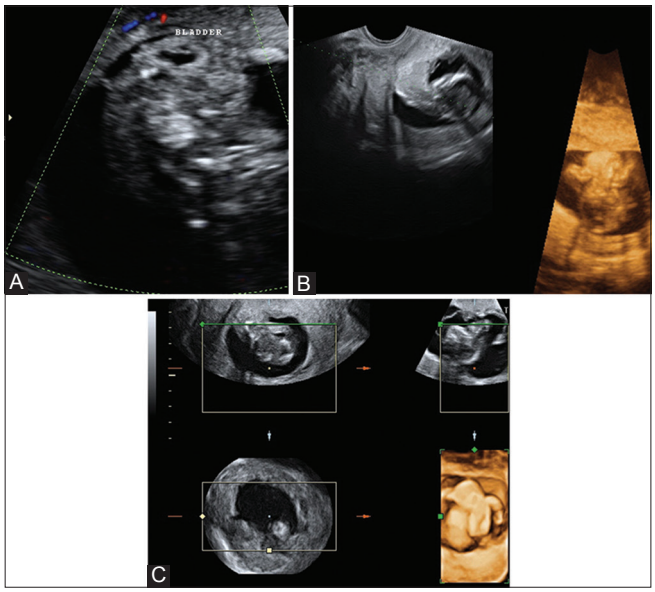

Figure 2 (A-C): (A) Gray-scale image showing intrafetal location of bladder. (B) Ultrasound image of fetus with kyphoscoliotic spine. (C) Three-dimensional ultrasound image showing grossly malformed fetus

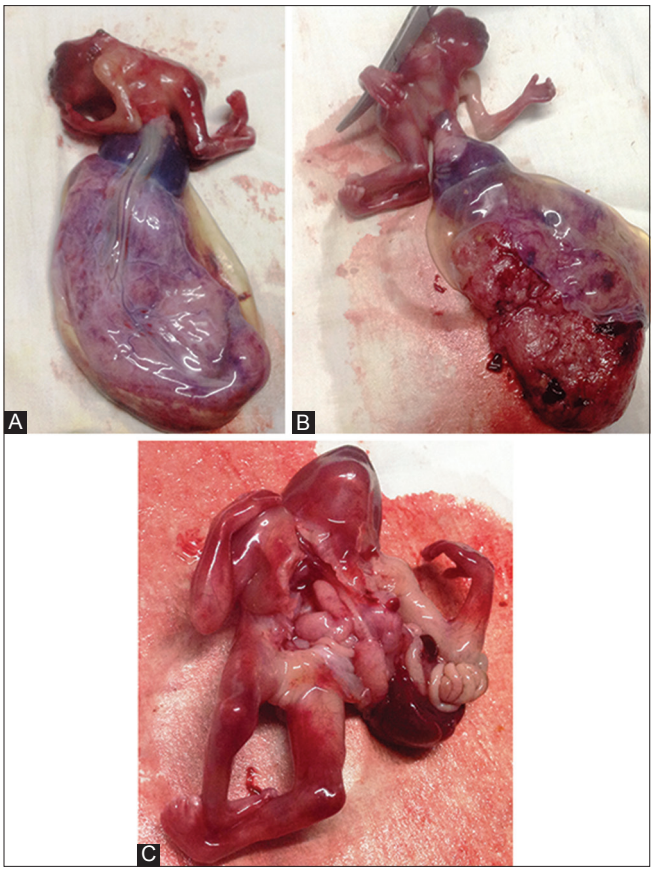

Figure 3 (A-C): (A) Fetus with short umbilical cord attached to the placenta with bilateral talipes. (B) Fetus with herniated organs covered by amniotic membrane. (C) Fetus after autopsy revealing liver and bowel loops within herniated sac

meningoceles, and placental anomalies characterized by the presence of short cord, persistence of extraembryonic coelom, and intact amnion.[11,12]

Antenatal USG in the second trimester not only confirms the diagnosis but also differentiates it from the other non-lethal dysplasias.

Other polymalformation complexes such as complex pentalogy of Cantrell, omphalocele-exostrophy-imperforate anus-spinal defects, and isolated gastroschisis should also be included in the differential diagnosis. The diagnosis of complete pentalogy of Cantrell requires demonstration of a supraumbilical wall defect, a lower sternal defect, a deficiency of the anterior diaphragm and the diaphragmatic pericardium, as well as an intracardiac defect.

USG features of cloacal exostrophy and the more severe omphalocele–exostrophy–imperforate anus–spinal defects complex include an absent fetal bladder, an infraumbilical abdominal wall defect, spinal distortion, and a sacral myelomeningocele.[13] In these conditions, the herniated abdominal wall contents are covered by a membrane and are not directly attached to the placenta, as occurs in body stalk.[14]

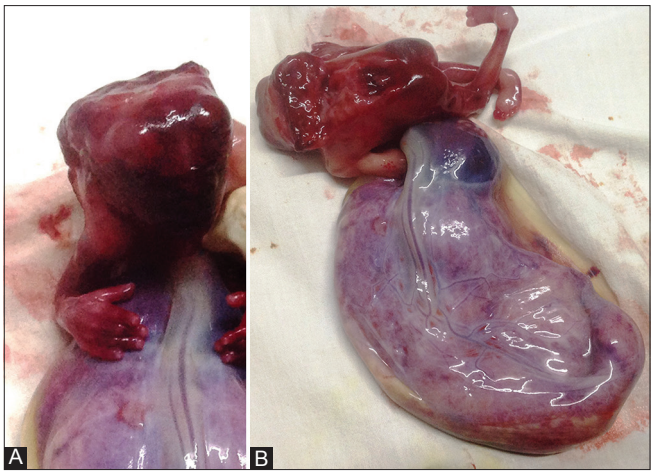

Figure 4 (A and B): (A) Fetus with anencephaly. (B) Fetus with open spina bifida with large defect

Low omphalocele is associated with bladder or cloacal exostrophy and caudal abnormalities such as meningomyelocele and anal atresia.

Gastroschisis is a congenital defect of anterior abdominal wall, usually to the right of the umbilical cord insertion. Gastroschisis is thought to result from an ischemic insult to the developing body wall.Intestines usually herniate through the defect in the anterior abdominal wall. However, the contents are not covered by amniotic membrane. Gastroschisis is usually an isolated anomaly and bears better prognosis.[15] The prognosis of body stalk complex is very poor compared to isolated exomphalos or gastroschisis.

Pregnancy should be terminated on establishing correct diagnosis, which requires careful ultrasound of the fetus whenever ventral abdominal wall defect is suspected.

The diagnosis of exomphalos should lead to search for other associated defects and chromosomal analysis should be considered. Isolated gastroschisis is the most benign of all. Thus, it is important to distinguish between these entities, as it will influence the choice of treatment and survival. The typical features can be detected ultrasonographically by the end of first trimester. Prenatal diagnosis of this anomaly would permit the early termination of pregnancy or avoidance of surgical intervention.

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Cite this article as: Kocherla K, Kumari V, Kocherla PR. Prenatal diagnosis of body stalk complex: A rare entity and review of literature. Indian J Radiol Imaging 2015;25:67-70.

Source of Support: Nil, **Conflict of Interest:** None declared.