CASE REPORT

A Case of Body Stalk Anomaly Arising after *Invitro* Fertilization at '26 week' Gestation

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ABSTRACT

Limb-body wall complex (LBWC) is a rare fatal congenital malformation syndrome. We describe a case of body stalk anomaly which was diagnosed at 26 weeks of primigravida patient with a history of primary infertility and had *in vitro* fertilization due to female factor. The fetus displayed multiple anomalies characteristic of body stalk syndrome including abdominal wall defect, kyphoscoliosis, and gross deformities of the lower limbs, which was diagnosed by ultrasound as early scan.

Key words: Body stalk anomaly, limb-body complex, multiple congenital anomalies, short cord syndrome, termination of pregnancy

INTRODUCTION

Body stalk anomaly or limb-body wall complex is a rare condition which accounts for about 0.32 per 100,000 births.[1] Despite its rarity, it has been reported in many case reports.[2]

Body stalk anomaly is usually characterized by the presence of a major anterior body wall defect, limb deformities, kyphoscoliosis, an absent or short umbilical cord, and/or craniofacial defects. Various other fetal abnormalities in which the abdominal organs develop outside of the abdominal cavity and remain attached directly to the placenta have also been reported.[3]

The sonographic landmarks include neural tube abnormalities, severe scoliosis, positional deformities, and abnormalities of fetal membranes. Various other fetal abnormalities might be present and maternal serum AFP levels are increased. Usually, fetuses with body stalk anomaly have a normal karyotype.[2]

This anomaly may occur in conjunction with defects of the neural tube, genitourinary malformations, intestinal atresia, and various anomalies of the chest wall and craniofacial defects, which led to the creation of a confusing range of terms for this disorder.[3]

Ultrasound diagnosis is very important. Diagnosis is based on whether two or three of mentioned abnormalities are present, and an early antenatal diagnosis is extremely important to enable the parents decide on options of continuation or termination of pregnancy.[4]

CASE PRESENTATION

A 31-year-old primigravida, not known case of any medical or surgical illness, was following as case of antenatal, no family history of any anomaly. She was not related to her husband and had *in vitro* fertilization due to female factor. The embryo transfer occurred on July 3, 2019, and the estimated date of delivery was April 27, 2020. She was first diagnosed at 26 + 4 weeks of gestation by ultrasound scan performed in a private clinic showing multiple fetal anomalies and scan was done with difficulty due to maternal habitus. By using two and three dimensional ultrasound revealed the fetus was stuck to

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the anterior uterine wall, placenta seen bipartite. The smaller part is anterior and giving the origin of the umbilical cord with single artery. Abdominal organs were seen with difficulty due to ventral wall defect only one kidney, urinary bladder and liver were seen. The chest with almost no lung tissue. The heart was pulsating with no clear morphology. There were vertebral column anomalies with angulation on two sites. Lower limbs were seen in free movements, only left arm was seen. Ultrasound was repeated during antenatal visits at 28 weeks showed spine deformity and four chambered heart. Asymmetry of cavities was noted. Large part of the back was attached to the membranes most probably due to amniotic membranes band, very short umbilical cord, possible absent abdominal wall, and difficult examination of the kidneys with positive fetal heart [Figures 1 and 2].

Case was discussed in perinatal forum meeting between the obstetrics and gynecology department and the neonatology department, they recommended termination of pregnancy as the fetus is incompatible with life. The family was counseled regarding the fatal condition of the fetus and chose to continue pregnancy.

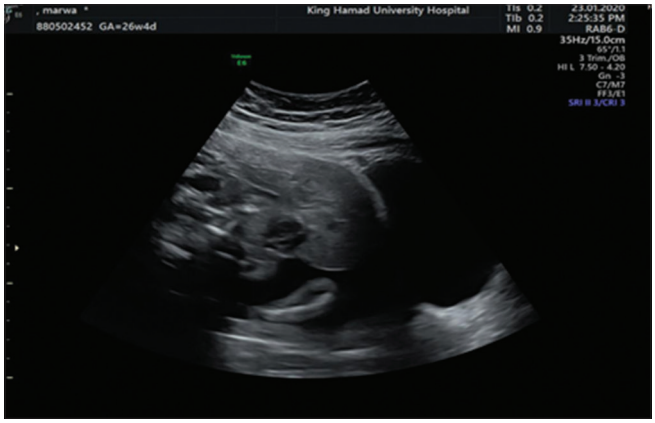

Figure 1: Ultrasound showing abdominal wall defect of body stalk anomaly at 26 weeks

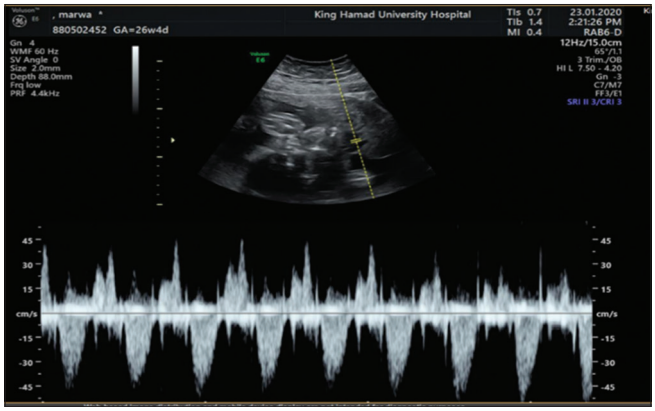

Figure 2: Ultrasound showing positive fetal heart beats at 26 weeks

At 32+3 weeks of gestation, the patient presented to the emergency department with labor pains as a result of transverse lie and was delivered by emergency cesarean section. The delivery was attended by neonatology consultant.

The newborn was male, weighing at 1.530 kg, with multiple congenital anomalies including abdominal wall defect, kyphoscoliosis, and gross deformities of the lower limbs, died immediately after birth and was sent to mortuary [Figure 3].

DISCUSSION

Body stalk anomalies belong to a group of massively disfiguring abdominal wall defects[5] which the abdominal, and often thoracic organs lie outside the body cavity but within a membrane composed of amnion and peritoneum. This sac is most often directly attached to the placenta.[5]

This case displayed typical features of body stalk anomaly in a fetus which was developing in the exocoelomic cavity.[5]

The reported prevalence has increased from approximately one in 14,000 pregnancies[1] to one in 7500 in more recent studies. This increase can be explained due to widespread use of ultrasound in the first trimester which enables an accurate diagnosis.[6]

This anomaly explained by three theories. The most accepted theory is an early rupture of the amnion before there is an obliteration of the coelom, but abdominal, spinal, and limb defects explained by passage of the lower part of the fetus into the extraembryonic coelom which resulting in an extreme form of the amniotic band sequence.[7]

Another theory explained that an abnormal folding of the trilaminar disc in its cephalic, caudal, and lateral directions that might lead to persistence of the coelomic cavity. As the embryo grows, the disk curves. It is transformed

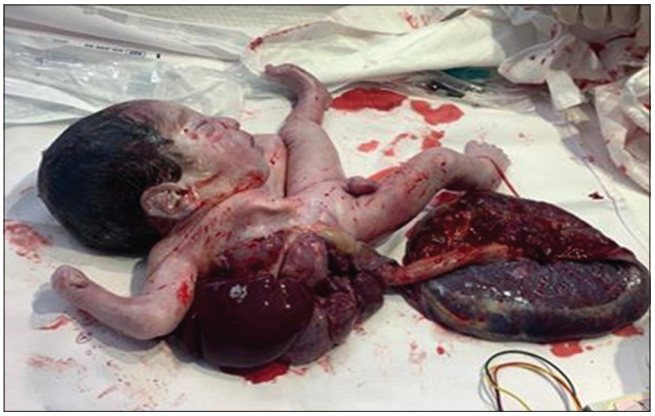

Figure 3: Body stalk anomaly of fetus after birth at 32 weeks of gestation

into a cylindrical foetus by circumferential folding. This process results in separation of the intraembryonic and extraembryonic cavities and formation of the body stalk (umbilical cord). The amniotic cavity, initially located dorsal to the germinal disk, grows rapidly to encircle the fetus and obliterate the extraembryonic coelom.[7]

Usually, fetuses with body stalk anomaly have a normal karyotype and therefore there is no need for routine karyotyping in these cases (2) which can be distinguished from other anterior abdominal and thoracic wall defects like omphalocele, gastroschisis, ectopia cordis, pentalogy of Cantrell, and OEIS.[7]

Omphalocele is characterized by supraumbilical defect with herniation of liver, bowel loops, and sometimes urinary bladder and herniated structures are covered by peritoneum. In gastroschisis, diagnosed when bowel loops floating in the amniotic cavity. OEIS includes many defects such as infraumbilical abdominal wall defect, exstrophy of bladder or absent bladder, imperforate anus and spinal dysraphism, and a sacral myelomeningocele. Pentalogy of Cantrell is a rare malformation characterized by lower sternal defects, anterior diaphragmatic defects, defect in the pericardium, cardiac ectopia, intracardiac anomalies, and a midline supraumbilical abdominal wall defect and all these anomalies related to sex chromosome and trisomy 13.[8]

In most of the described cases, the karyotypes of the affected fetuses are completely normal, and only in two cases there have been chromosomal abnormalities associated with uniparental disomy of chromosome 16 and with a trisomy of chromosome 2 due to a confined placental mosaicism. But what really known that body stalk anomaly related to environmental and genetic factors with no abnormality in karyotyping , still this complex poorly understood condition.[9]

CONCLUSION

This case report showed body stalk anomaly resulted from *in vitro* fertilization and diagnosed in the 2nd trimester. It is a lethal fetal anomaly, incompatible with life, however, it is very important to counsel the parents regarding the options of continuation or termination of pregnancy and to be reassured for non-recurrence of other lesions in future pregnancies.

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