Abdominal Wall Defect Found at the First-trimester Ultrasound Scan

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Section 2 – Answer

Case description

We report a case of a nulliparous 28-year-old Portuguese woman at 14 weeks of gestation for her first prenatal visit. She was healthy without known underlying conditions or surgical history.

First trimester combined screening revealed a reduced risk for trisomy 21 (1:4403), trisomy 18 (1:55517), and trisomy 13 (1:42660) with pregnancy-associated plasma protein-A = 0.58 MoM and free beta-human chorionic gonadotropin = 0.59 MoM.

In ultrasound (US), we found a live fetus with indeterminable crown-rump length (CRL) due to the inability of obtaining a full longitudinal view of the appropriate structures [Figures 1 and 2]. The nuchal translucency was impossible to measure [Figure 3].

The fetus appeared fixed in position. There was a severe midline defect of the fetal abdominal wall with a protruding abdominal mass adherent to the placenta suggesting an evisceration of the abdominal contents [Figure 4]. The cord insertion site could not be identified, and no free-floating loops of cord were seen. In a cross-section view of the abdomen, there was a marked deviation of misalignment of the lower limbs [Figure 5]. The placenta was located on the anterior wall of the uterus, and the amniotic fluid samount was normal. Cytogenetic study for aneuploidy revealed a normal karyotype. The postabortion study confirmed the US findings [Figures 6 and 7].

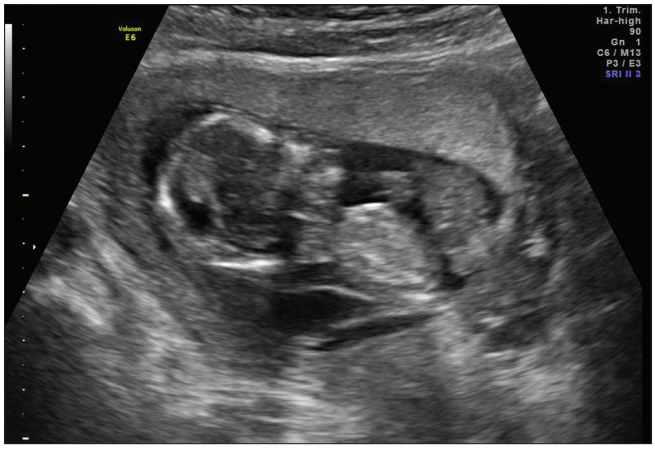

Figure 1: Ultrasound: Indeterminable crown-rump length

Interpretation

The US findings included a large abdominal defect, abnormalities in the axial skeleton such as scoliosis, and a short or absent umbilical cord. Before establishing a final diagnosis, it is important to consider all pathologies that can affect the

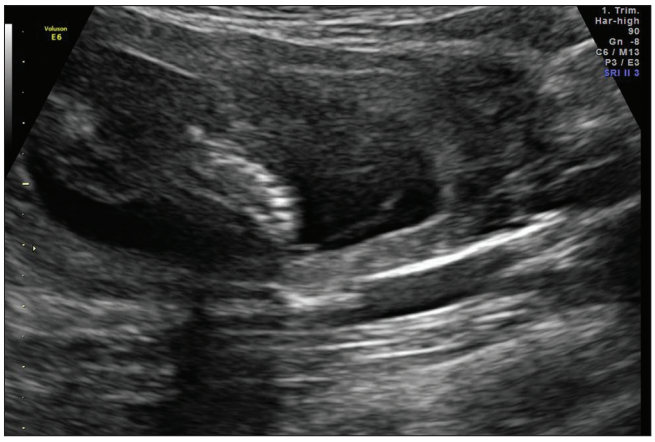

Figure 2: Ultrasound: Abnormal spine suggesting scoliosis

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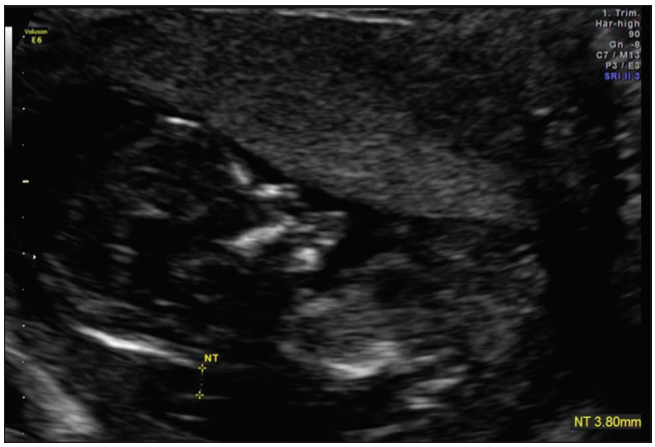

Figure 3: Ultrasound: Longitudinal view of the head

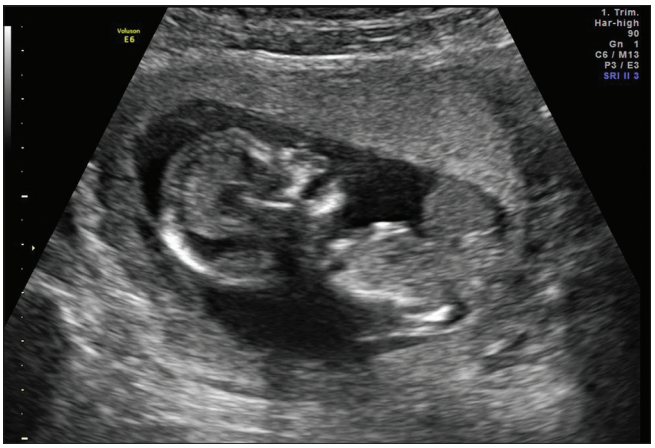

Figure 4: Ultrasound: Abdominal mass adherent to the placenta surface

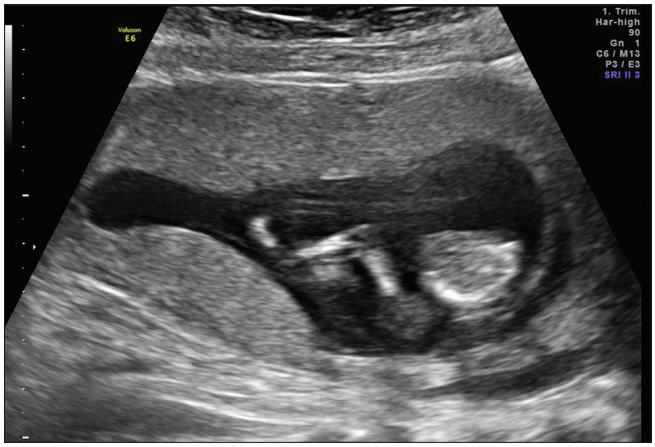

Figure 5: Ultrasound: Cross-section view of the abdomen-deviation of the lumbar spine and misaligned lower limbs

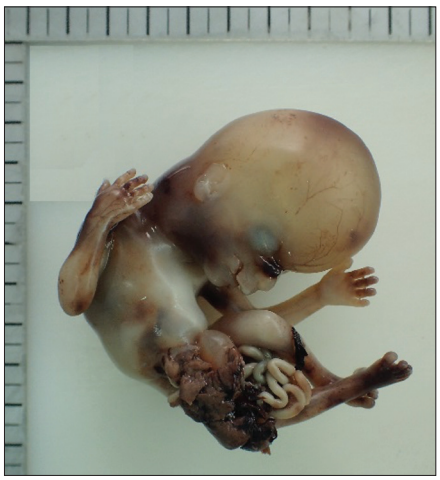

Figure 6: Postabortion images-severe kyphoscoliotic with asymmetric lower limbs and evisceration of the abdominal contents

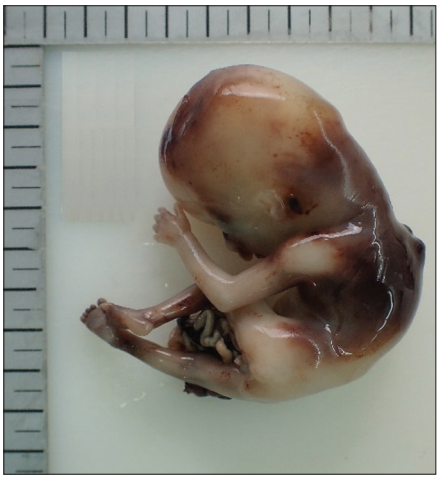

Figure 7: Postabortion images-severe kyphoscoliotic with asymmetric lower limbs and evisceration of the abdominal contents

abdominal wall integrity, such as omphalocele, gastroschisis, vesical exstrophy, Cantrell pentalogy, amniotic band syndrome, Beckwith–Wiedemann syndrome, body stalk anomaly, and the OEIS complex (omphalocele, exstrophy of cloaca, imperforate anus, and spinal defects).

The patient was referred to our prenatal diagnosis department for antenatal counseling and further investigation. At 14 weeks, the patient underwent chorionic villus sampling. The cytogenetic examination revealed a karyotype 46 × X and a normal molecular study by Array-comparative genomic hybridization.

The postabortion study confirmed the US findings. The fetus had a large ventral abdominal wall defect. An abdominal mass containing the liver, spleen, and bowel loops was seen herniating through the abdominal wall defect, and intrathoracic protrusion of retroperitoneal organs was confirmed. Skeletal deformities were severe kyphoscoliotic and asymmetric lower limbs. A short umbilical cord was also confirmed.

Given these findings, the diagnosis was consistent with the limb-body wall complex.

Body stalk anomaly is a rare malformation syndrome with a reported incidence of 1/14,000–1/31,000 pregnancies in large epidemiologic studies.[1] It is defined by a severe defect of the abdominal wall, in which there is evisceration of abdominal organs and in more severe cases of thoracic organs as well.

This congenital malformation is accompanied by severe kyphoscoliosis and the presence of a rudimentary umbilical cord which is usually short or even absent.[2]

An appropriate midsagittal view of the fetus for the measurement of the CRL, and adequate sweeps through the head and abdomen, should identify all the cases of body stalk anomaly between 11 and 13 weeks of gestation.[3]

The exact pathophysiology and trigger factors are still unknown. Much remains to be elucidated in terms of its real epidemiology, global distribution, and risk factors. In addition, efforts should focus on making an early diagnosis to provide the expecting parents the necessary information and counseling regarding the prognosis of the anomaly. Special emphasis on the benefits of early management and possible complications that can arise during childbirth and/or pregnancy should be placed. The high mortality rate of the anomaly should also be informed.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initial will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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