

Skeletal Defect at First-trimester Ultrasound Scan

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SECTION 2 – ANSWER

Case description

We report the case of a nulliparous 36-year-old Portuguese female at 14 weeks of gestation for her first prenatal visit in our hospital. The medical history was unremarkable without known underlying conditions (medical or surgical).

In ultrasound (US) performed at 14 weeks and 3 days, we found a singleton live fetus with normal amniotic fluid volume. Fetal biometry was not consistent with late period date because the crown-rump-length was found to be smaller than expected. The fetal anatomy above the midthorax, including the heart and the intracranial structures, appeared normal. Below this level, however, a sudden termination of spine at the lumbosacral level was seen [Figure 1]. In the transverse section, the sacrum was missing, and the iliac wings are positioned in midline close to each other [Figure 2].

The lower limbs were akinetic and were kept in the fixed flexion [Figures 3 and 4]. The fetus had club feet [Figure 5].



Figure 1: Ultrasound: Abrupt termination of spine at lumbosacral level

The bladder was not observed, and the bowel was hyperechogenic [Figure 6].

First trimester combined screening was performed and revealed a reduced risk for trisomy 21 (1:13,494), trisomy 18 (1:91,068), and trisomy 13 (1:76,419) with pregnancy-associated plasma protein-A = 0.68 MoM and free beta-human chorionic gonadotropin = 0.60 MoM.

Taking into account the US findings, a cytogenetic study was proposed, which revealed a normal array-comparative genomic hybridization. The postabortion study confirmed the US findings.

INTERPRETATION

The US findings included an absence of the lumbosacral spine, flexion contractures of the lower extremities, club feet, and hyperechogenic bowel. Before establishing a final diagnosis, it is important to consider all pathologies with caudal defects such

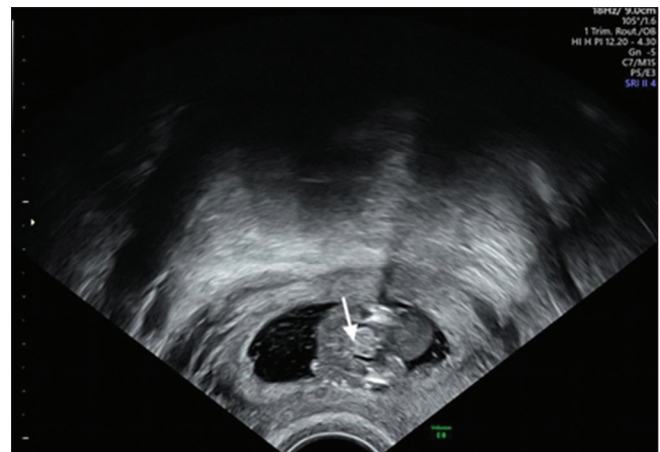


Figure 2: Ultrasound: Missing sacrum

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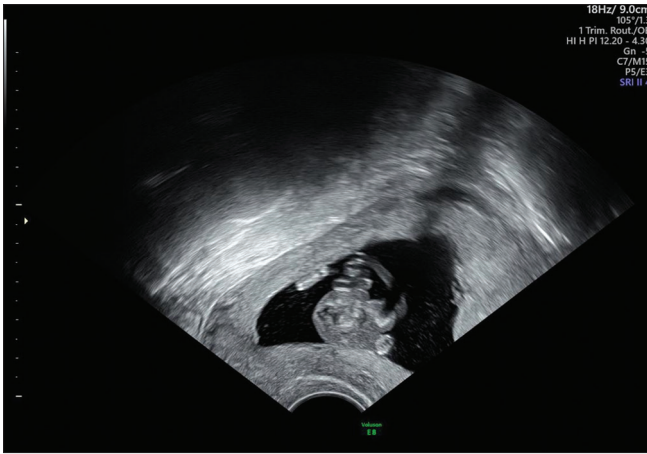


Figure 3: Ultrasound: Transversal view - Fixed flexion of lower limbs 15

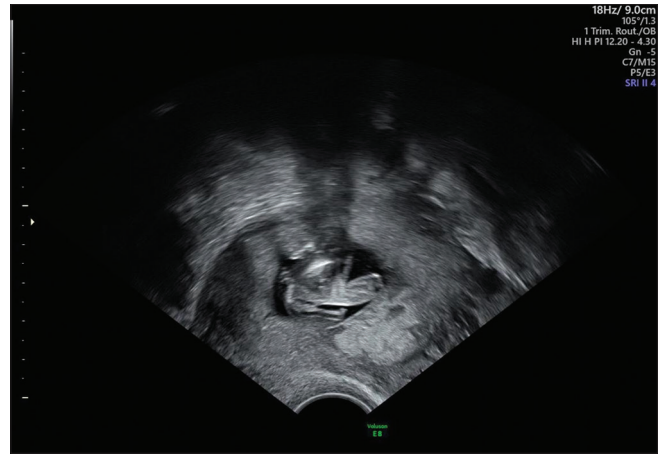


Figure 4: Ultrasound: Fixed flexion of lower limbs

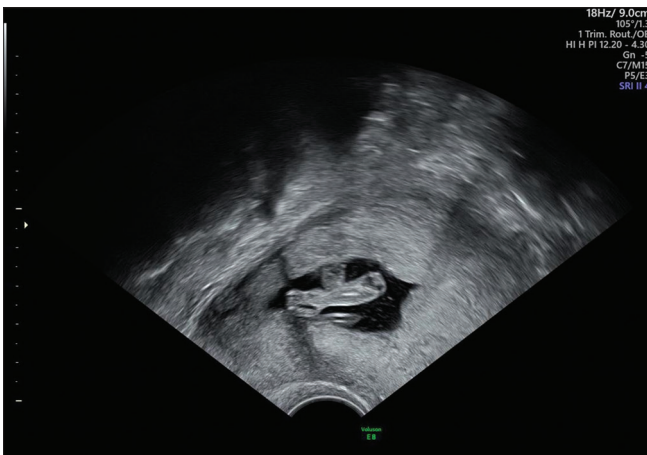


Figure 5: Ultrasound: Club feet

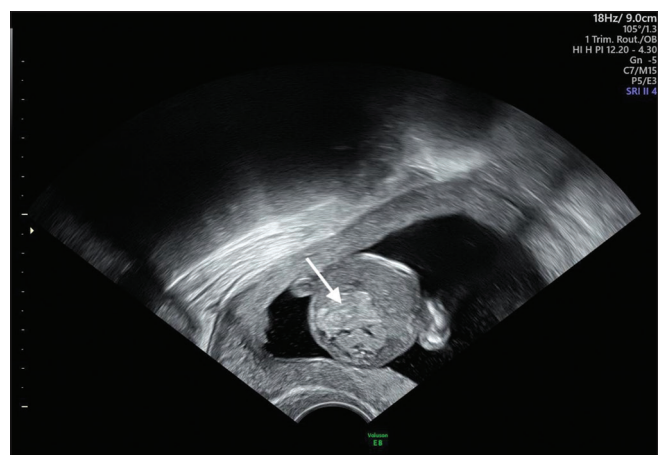


Figure 6: Ultrasound: Hyperecogenic bowel

as body-wall complex and segmental spinal dysgenesis (SSD), vertebral, anorectal, cardiac, tracheoesophageal, renal and limb (VACTERL) complex, Currarino syndrome (CS), and syringomyelia. Bladder exstrophy, omphalocele, and sacral myelomeningocele can be demonstrated in the body-wall complex. SSD is a rare congenital abnormality in which a segment of the spine and spinal cord fails to develop properly.^[1] Severe forms of the disease are commonly associated with cardiac, renal, and respiratory problems with overlapping feature of VACTERL complex (anomalies). CS is characterized by the triad of anorectal malformations, sacral bone defects, and presacral mass and in sirenómelia we had complete or partial fusion of lower limbs.

This patient was referred to our prenatal diagnosis department for antenatal counseling and further investigation. At 15 weeks, the patient underwent amniocentesis. The cytogenetic examination revealed a karyotype 46 XY and a normal molecular study by Array-comparative genomic hybridization. The patient was counseled accordingly, and she elected for the termination of pregnancy.

Postabortion study confirmed the prenatal diagnosis, showing absent lumbosacral vertebrae, pelvic deformities, and femoral hypoplasia. Muscular atrophy of the lower extremities with

extensive popliteal webbing and club feet was confirmed. The fetus had also urinary anomalies, with an isolated horseshoe kidney in the midline and collapsed bladder.

Given these findings, the diagnosis was consistent with a caudal regression syndrome.

Caudal regression is a rare syndrome which has a spectrum of congenital malformations involving approximately 1 in 25,000 live births ranging from simple anal atresia to absence of sacral, lumbar and possibly lower thoracic vertebrae, to the most severe form which is known as sirenómelia.^[2]

This congenital abnormality includes agenesis of the lumbar spine, sacrum, and coccyx, as well as hypoplasia of the lower extremities. It is considered the most characteristic of all congenital anomalies associated with maternal diabetes mellitus.^[3] Orthopedic, genitourinary, gastrointestinal, and cardiac anomalies can be associated.^[4] In our case, club feet, flexion contractures of hips and knees were the orthopedic deformities, and we had also urinary anomalies.

The embryologic insult occurs at the midposterior axis mesoderm, and the lesion originates before the 4th weeks of gestation, but the etiology remains unclear.^[5]

The prognosis is good when isolated sacral agenesis, but can be worse if associated with others abnormalities.

The diagnosis is done by US, but can be difficult before 19 weeks because the sacrum is not enough calcified.^[6]

Detailed evaluation of the fetal spine and lower extremities is an important aspect of every prenatal US examination.

In the presence of normal amniotic fluid, the diagnosis can be made by demonstrating the termination of lumbar spine and small and abnormal lower extremities.^[3] Both findings were present in this clinical case.

Furthermore, the detection of the abnormality was at 14 weeks of pregnancy. Early antenatal sonographic diagnosis is important in view of the dismal prognosis and allows for earlier and less traumatic termination of pregnancy.

Declaration of patient consent

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

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Nil.

Conflicts of interest

There are no conflicts of interest.

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