

## Skeletal Defect at First-trimester Ultrasound Scan

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### SECTION 1 – QUIZ

#### Case description

We report a case of a nulliparous 36-year-old Portuguese woman 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 the spine at 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 fixed flexion [Figures 3 and 4]. The fetus had clubfeet [Figure 5]. The bladder was not observed and the bowel was hyperechogenic [Figure 6].

Combined first-trimester screening was performed and revealed a reduced risk for trisomy 21 (1:13494), trisomy 18 (1:91068), and trisomy 13 (1:76419) 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.

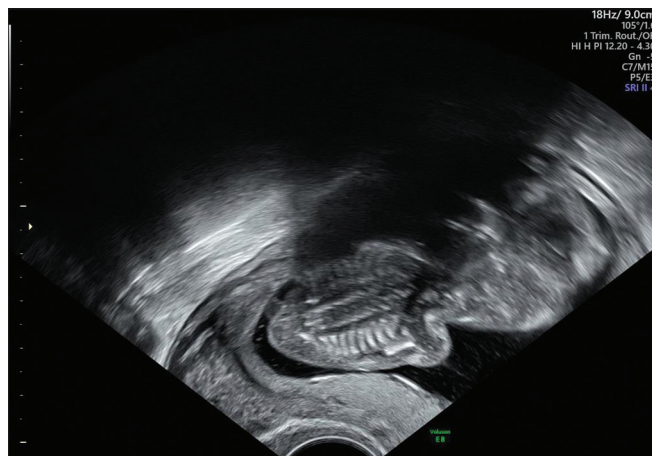


Figure 1: Ultrasound: Abrupt termination of spine at lumbosacral level



Figure 2: Ultrasound: Missing sacrum

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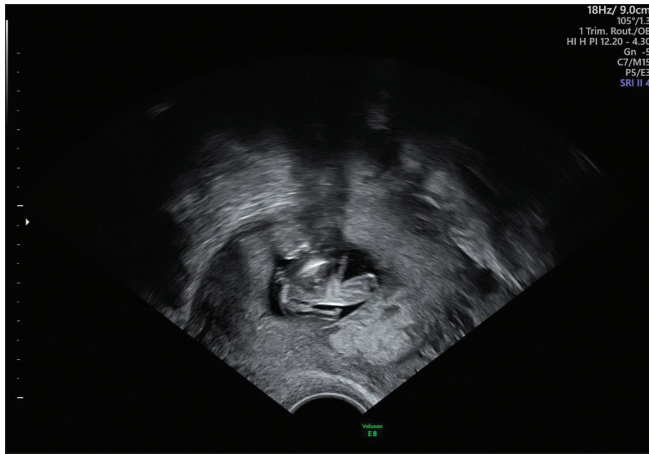
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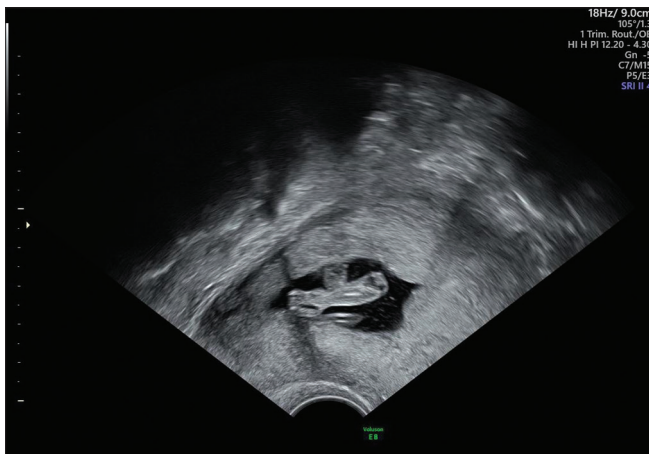
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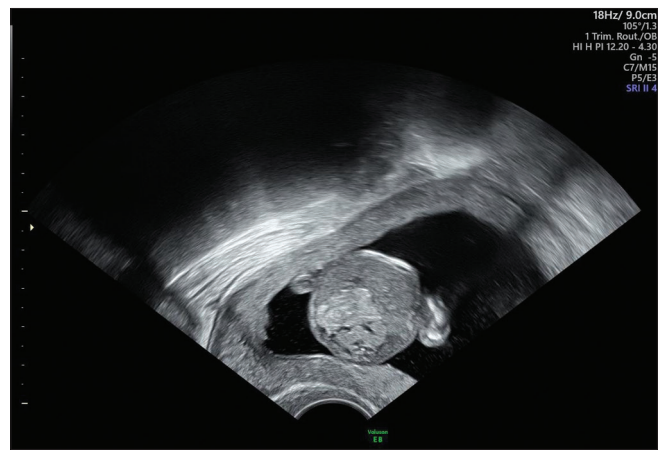
**Figure 3:** Ultrasound: Transversal view - Fixed flexion of lower limbs 15



**Figure 4:** Ultrasound: Fixed flexion of lower limbs



**Figure 5:** Ultrasound: Clubfeet



**Figure 6:** Ultrasound: Hyperechogenic bowel

## WHAT IS THE DIAGNOSIS?

### 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.

### Financial support and sponsorship

Nil.

### Conflicts of interest

There are no conflicts of interest.