

Spine Abnormality in a Fetus at 21 Weeks of Gestation

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SECTION 1 – QUIZ

Case description

A 29-year-old pregnant woman, 2 gravida 1 para (one cesarean section 3 years ago, mother of a healthy child) was sent to our hospital at 13 weeks of gestation because of the personal history of Type 1 diabetes mellitus diagnosed at 25-year-old. She was medicated with long-acting insulin achieving adequate glycemic control.

First trimester combined screening revealed a low risk for aneuploidies: Trisomy 21 (1:13809), 18 (1:2282), and 13 (1:1733). The ultrasound showed a live fetus with a cephalocaudal length of 54.7 mm (gestational age of 12 weeks), nuchal translucency of 1.6 mm (<95th centile for gestational age), and present nasal bone. The skull, brain, spine, heart, abdominal wall, stomach, bladder, both hands, and feet were scanned, and no abnormalities were found. The amniotic fluid was normal.

The second-trimester ultrasonography, performed at 21 weeks and 5 days of gestation, showed a live fetus in the breech position. The fetus had a head circumference of 189.9 mm and a biparietal diameter of 53.4 mm, both in

the 50th centile for gestational age. The skull had a normal shape with no intracranial abnormalities, including cystic formations or ventricular dilation. The face had no apparent dysmorphism. Evaluation of facial profile showed nasal bone hypoplasia (4.4 mm) [Figure 1]. Cross-section view of the neck had nuchal edema (6.5 mm) [Figure 2].

The thoracic evaluation showed no apparent abnormalities, including both lungs, heart, and mediastinum. Abdominal circumference was 182.1 (centile 50–75th for gestational age). The stomach, both kidneys, bladder, and external male genitalia were normal.

The spinal sagittal section showed signs of deviation with an apparent hemivertebra and thoracolumbar scoliosis [Figures 3 and 4]. The limbs and extremities had normal structure and shape; long bones were short for gestational age with a humerus of 29.2 mm, the radius of 24.8 mm, ulna of 27.5 mm, femur of 29.3 mm, tibia of 24.6 mm and fibula of

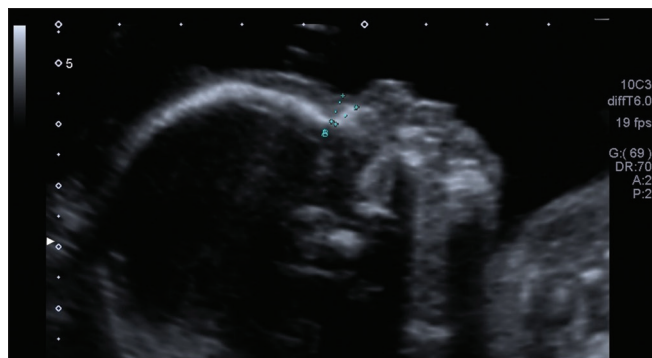


Figure 1: Ultrasound: Fetal facial profile showing nasal bone hypoplasia

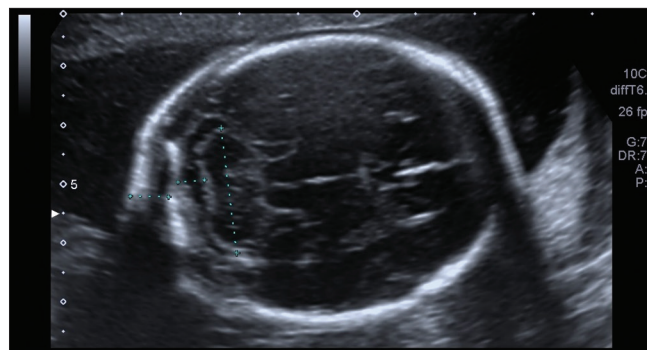


Figure 2: Ultrasound: Cross-section view of the neck showing nuchal edema

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Figure 3: Ultrasound: Sagittal section of the spine showing deviation with an apparent hemivertebra

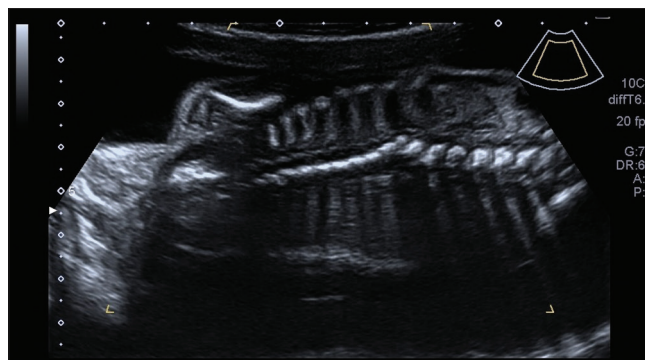


Figure 4: Ultrasound: Sagittal section of the spine showing deviation with an apparent hemivertebra

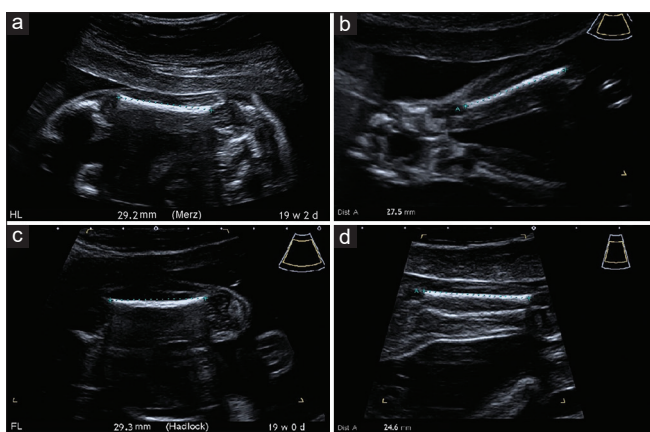


Figure 5: Ultrasound: Shortening of the long bones (below 5th centile). No apparent abnormalities (a-humerus; b-ulna; c-femur; d-tibia)

25.4 mm, all below the 5th centile [Figure 5]. No bone fractures were identified.

The placenta was located on the posterior wall of the uterus, and the amniotic fluid was normal.

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 initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.