A Pattern of Malformations in the First Trimester Ultrasound

Sara Sereno[1]*, Sofia Pedrosa de Moura[1], Matilde Martins[1], Inês Falcão Reis[1], Rosete Nogueira[2], Célia Araújo[1]

[1]Department of Obstetrics and Gynecology, Local Health Unit Entre o Douro e Vouga, Santa Maria da Feira, Portugal, [2]Department of Embriofetal and Placentar Pathology, Unilabs – CGC Genetics (Clinical Genetics Center), Porto, Portugal

Section 1 – Quiz

CASE

A 33-year-old woman, primigravida, with a medical history of euthyroid autoimmune thyroiditis, was referred to our prenatal diagnostic center due to suspicion of multiple malformations detected during an ultrasound performed at her obstetric appointment at 11 weeks of gestational age. The obstetric ultrasound showed a thickened nuchal translucency, an encephalocele [Figure 1] and a midline defect including an omphalocele and ectopia cordis [Figures 2 and 3].

Chorionic villus sampling was performed. Genetic analysis revealed a normal karyotype (46, XY) and array of CGH findings.

What is your initial diagnostic hypothesis?

Ethics statement

This study was conducted in accordance with the ethical principles outlined in the Declaration of Helsinki and its amendments. 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 The patient understand that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.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.

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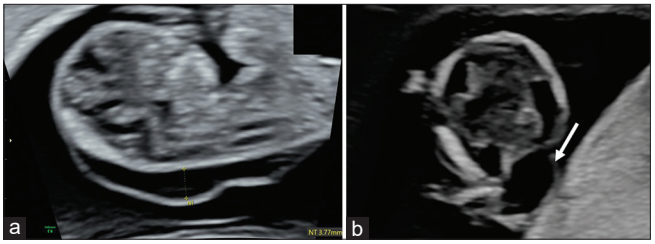

Figure 1: Obstetric ultrasound showing a thickened nuchal translucency in a sagittal section (a) and an encephalocele (b, full arrow) in a transverse section of the fetal head

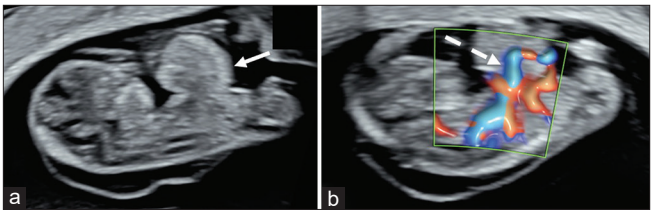

Figure 2: Obstetric ultrasound: midline sagittal section of the fetal body showing a midline defect with omphalocele (a, full arrow) and ectopia cordis (b, dashed arrow), as evidenced with the use of colour Doppler

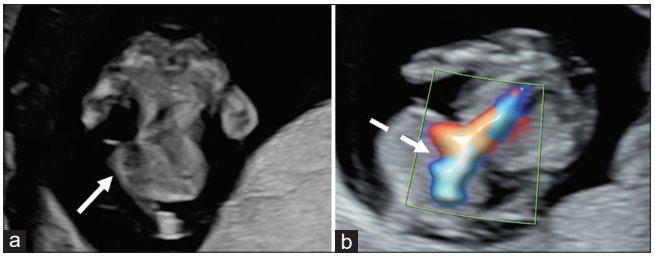

Figure 3: Obstetric ultrasound: transverse section of the midline defect (a, full arrow) and ectopia cordis (b, dashed arrow) without (a) and with (b) color Doppler

Address for correspondence: Dr. Sara Sereno, Department of Obstetrics and Gynecology, Local Health Unit Entre o Douro e Vouga, Hospital de São Sebastião, Rua Doutor Cândido Pinho, 4520-220, Santa Maria da Feira, Portugal.
E-mail: sarasereno13@gmail.com

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