Prenatal diagnosis of fetal hemivertebra at 12 weeks of gestation

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DESCRIPTION

A pregnant woman in her later 30s, with a history of a previous uneventful pregnancy with full-term delivery of a healthy baby and subsequent four first trimester miscarriages, all with the same partner. The study for recurrent early pregnancy loss was negative (karyotype, hormonal disorders, thrombophilia screening, endometrial assessment) and the couple's family histories were unremarkable. The woman was previously healthy, with folic acid supplementation preconceptionally and with no history of exposure to teratogens in the first trimester.

First trimester ultrasound revealed a singleton pregnancy, with crown-rump length compatible with 12 weeks' gestation and no sonographic markers of common foetal trisomies. During morphological examination, a slight distortion of the fetal column at thoracic level was evident in the coronal view, making the diagnosis of a fetal scoliosis (figure 1). No other abnormalities were found. Follow-up ultrasound examination at 20 weeks of gestation showed a more evident thoracic scoliosis and a small triangular body was seen wedged against the normal vertebral bodies, making the diagnosis of thoracic hemivertebra. No further morphological anomalies were found.

Amniocentesis was performed, demonstrating normal fetal female karyotype and normal array-CGH (Comparative Genomic Hybridization). Due to the progression of spine angulation at thoracic level (associated with poor prognosis), after multidisciplinary counselling, the couple elected to terminate the pregnancy.

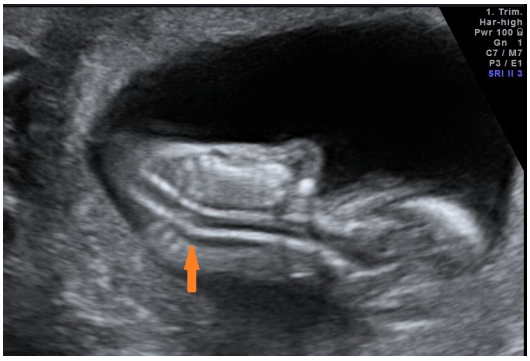

Figure 1 First trimester evaluation: arrow shows distortion of the fetal column at thoracic level (coronal view).

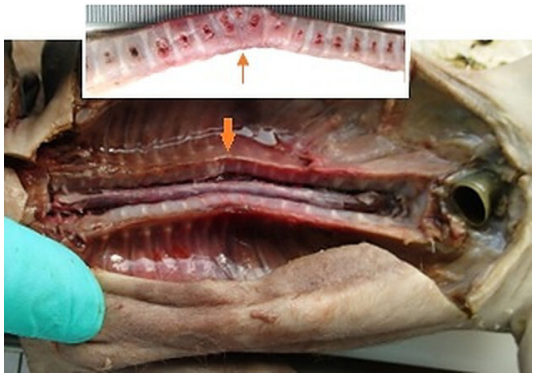

Figure 2 Postmortem examination: a thoracic hemivertebra with a collapsed contiguous vertebra (arrow).

Postmortem examination revealed a female fetus with an isolated skeletal malformation: a thoracic hemivertebra with a collapsed contiguous vertebra and absence of the right rib, which was an unsurprising finding, due to its thoracic origin (figure 2).

Hemivertebra is an uncommon congenital vertebral abnormality, in which only half of the vertebra develops, occurring in 0.5–1 per 1000 births.[1 2] Aetiology is unknown. Hypotheses include an abnormal distribution of the intersegmental arteries of the vertebral column, sclerotomal defect, poor nutritional intake and genetic syndromes including *Jarcho-Levin*, *Klipplel-Fiel* and *VACTERL* (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal and limb anomalies).[2] Although most cases present with coexisting anomalies, situations of isolated hemivertebra seem to be simple structural defects, unaccompanied by genetic damage or with environmental causes.[1–3]

The hemivertebra acts as a wedge within the vertebral column, causing contralateral deviation of the spine at the level of the abnormal vertebra, causing scoliosis, lordosis or kyphosis.[1 3] This can be observed prenatally by 2-dimensional ultrasound, which shows a triangular-shaped ossified structure wedged on the vertebral column and the resulting spinal curvature. Due to the size of the fetal spine and the poor ossification in the first trimester of pregnancy, the diagnosis, if achieved prenatally, usually occurs in the second trimester.[1–5] Studies suggest that the earlier the diagnosis, the higher likelihood of severe spinal abnormality, higher

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incidence of coexisting abnormalities and worse outcome.[2] Prognosis is also closely related to the site of occurrence. Cervical or thoracic hemivertebra are associated with a higher risk of spinal cord abnormalities and, therefore, with a poorer outcome.[1] Surgical resection of the affected segment followed by posterior spinal fusion is the most common therapy for hemivertebra and has had promising results.[6–8]

Learning points

- When prenatal diagnosis is achieved, it usually occurs in the second trimester of pregnancy, with a minority of cases reported in the first trimester of pregnancy. In this case, the hemivertebra was suspected at 12 weeks' gestation due to early presentation fetal scoliosis.
- Prenatal diagnosis of hemivertebra is feasible in the first trimester of pregnancy, providing the parents important information and support to make an informed decision early in pregnancy or early postnatal referral for care and treatment.
- Therefore, a thorough inspection of the vertebral column should be made systematically during the first trimester ultrasound.

Contributors DM and ISG were directly responsible for the initial approach to the patient. Follow-up of the patient and pregnancy termination were oriented by ISG and CG. Postmortem evaluation was made by RNN. All authors were major contributors in writing the manuscript. All authors read and approved the final manuscript.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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