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Running Head

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ABSTRACT

We herein present a case of fetal multiple hemivertebrae detected at antenatal sonography. The use of the 3D technology supported by a new contrast enhancement rendering algorithm (Crystal Vue) has allowed the accurate prenatal classification of the defect, confirmed at follow up, that would have been difficult to define by 2D only.

REPORT

A 30-years G1 Caucasian was referred at 20 weeks to the ultrasound laboratory of our Department of Fetal Medicine for a dedicated anomaly scan, due to a nuchal translucency above the 99th centile at 12 weeks of gestation (3.7 mm). Fetal karyotype was normal after chorionic villous sampling. The patient's medical and obstetric history was unremarkable.

At 2D ultrasound scan an abnormal lateral deviation of the fetal spine was noted on the coronal plane at the level of the thorax. The skin overlying the spine appeared intact, and there were no obvious signs of associated anomalies.

A 3D volume of the fetal trunk was acquired sagittally and reconstructed on the coronal plane using maximum mode and the new contrast enhancement rendering algorithm Crystal Vue (Samsung WS 80 Elite system), documenting multiple and fused hemivertebrae at the level of the thoracic (T3-4, T7 and T10 with absence of the corresponding ribs) and of the lumbar spine (L1-L2). (Figure 1)

After detailed counselling with the Geneticist and the Paediatric Orthopaedic Surgeon the couple opted for the termination of pregnancy.

The antenatal ultrasound findings were confirmed post mortem at X Ray (Figure 1)

Haemivertebra is a congenital abnormality of the spine, which occurs in 1-10 per thousand births[1]. It is believed to result from the failure of one of the lateral chondrification centres to develop, which causes the absence of half of the vertebral body, the corresponding

rib and neural arch[1]. The aetiology is unknown but it has been suggested that abnormal distribution of intersegmental arteries of the vertebral column may be involved in the pathogenesis[1]. The defect may affect single or multiple vertebrae causing congenital scoliosis[1]. A haemivertebra may be associated with other structural defects, particularly those of the musculoskeletal system. Cardiac and genitourinary tract anomalies are the most common extra-musculoskeletal anomalies associated with hemivertebra. An association between increased nuchal translucency (> 3.5 mm) in the first trimester and kyphoscoliosis has also been reported[2].

The incidence of chromosomal abnormalities in cases of isolated hemivertebra is reported to be small as described by Zelop et al. (normal karyotype in 18 fetuses with isolated finding). Although an underlying chromosomal abnormality is uncommon, hemivertebrae especially when multiple, may be part of a genetic syndrome such as Jarcho-Levin syndrome, Klipper-Feil syndrome and VA(C)TER(L) [3].

The prognosis of isolated single hemivertebra is usually excellent with only a few cases requiring postnatal surgery or physical therapy for congenital scoliosis or functional impairment[4]. In case of multiple hemivertebrae the outcome is dependent on the site and the number of the involved segments[4] with a possible risk of spinal cord involvement and mechanical respiratory impairment if cervical and thoracic spine are extensively affected. Overall, if untreated, about 25% of patients show no progression of the scoliosis, in 50% the spine abnormality progresses slowly and in the remaining 25% there is a rapid worsening[4].

There are a few reports of fetal hemivertebra diagnosed antenatally by means of 2D ultrasound[5]. In these cases, the 2D sagittal section of the spine, which is the standard view to evaluate its integrity, may be unremarkable (figure 2), while the coronal view of the spine is able to spot both the vertebral anomaly and the scoliosis when associated.

Hemivertebra may have a similar ultrasonic appearance of other vertebral anomalies, which cause congenital scoliosis including wedge vertebra, butterfly vertebra, bloc

vertebra, bar vertebra. Differential diagnosis often requires a neonatal radiologic evaluation. Neural tube defects may also cause an abnormal curvature of the spine, but their association with intracranial changes as well as the presence of a meningocele/myelomeningocele sac may help in the differential diagnosis. Eventually diastematomyelia may also have a similar sonographic appearance, but in these cases a peculiar image of a vertebra with three posterior ossification centres may be obtained in axial view.

The introduction of 3D ultrasound in fetal imaging has allowed a better antenatal characterization of the haemivertebra thanks to the volume reconstruction of the spine on the coronal plane (Figure 1, 2).

The 3D ultrasound of fetal spine may be extremely helpful when dealing with the antenatal suspicion of fetal spine anomalies such as haemivertebra. This technology supported by the use of new dedicated software allows to obtain an accurate, fast and reproducible diagnosis of the defect at prenatal ultrasound.

The use of new contrast enhancement rendering algorithm such as Crystal Vue seems to be particularly accurate in the imaging of the fetal spine and ribs. In our case of fetal multiple haemivertebra, 3D ultrasound imaging supported by Cristal Vue enabled the precise characterization of the defect, in terms of number and location of the vertebral bodies involved. On this basis, the more relevant prognostic aspects have been derived to counsel the prospective parents.

DISCLOSURE STATEMENT

None of the authors reports any conflicts of interest. No funding was received. The subjects involved have given informed consent.

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