

Ultrasound and autoptic diagnosis of asphyxiating thoracic dysplasia

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Background. The skeletal system develops from mesoderm. In most bones (e.g., the long bones), ossification is preceded by cartilage (endochondral ossification). In other cases, such as flat bones, ossification develops directly in the mesenchyme without cartilage formation (intramembranous ossification). Skeletal dysplasias are a heterogeneous group of more disorders associated with developmental abnormalities of bone and cartilage. The modes of transmission are similar: autosomal dominant and recessive and X-linked dominant and recessive. Despite the potential advantages of 3-dimensional ultrasound (3D-US), antenatal diagnosis of skeletal dysplasia is difficult, given the large variety and complexity of these disorders: their phenotypes are variables and their features are overlapping. We present a case report of a woman with prenatal diagnosis of skeletal thoracic dysplasia, confirmed by postnatal radiography and fetal autopsy.

Case report. A 26-year-old woman, primigravida, was referred for routine ultrasonic examination during her second trimester of pregnancy. Ultrasonography (USG) showed a single live foetus of a gestational age of 20+3 weeks; biparietal diameter and head circumference were adequate for the week of gestation. There was a polyhydramnios. The fetal thorax was extremely narrow. The thoracic circumference (TC) measured 100 mm (< 5th percentile), the abdominal circumference (AC) measured 157 mm (50th percentile), and the TC/AC ratio was 0.64 (normal range: 0.77–1.01). The long-bone lengths measured < 5th percentile, especially the proximal part of the upper limbs. Ultrasound scans of fetal abdomen revealed bilateral slight increase in the size of kidneys. There were no neural tube defects, and the fetal stomach and urinary bladder were normal. Fetal echocardiography revealed mild ventricular septal defect with good hemodynamic effect. Based on these findings, the diagnosis postulated as possible was asphyxiating thorax dysplasia (ATD). After genetic counseling, the patient decided for an elective termination of the pregnancy. A stillborn male fetus was delivered with a weight of 470 g. Infantogram and gross autopsy findings (narrow thorax, short upper limb bones, poor definition of pyramids of kidneys) supported the diagnosis made.

Conclusion. Although skeletal anomalies are difficult to diagnose antenatally, a detailed scan of fetal anatomy between 20 and 32 gestational weeks exclude majority of major skeletal dysplasias. Termination of pregnancy is indicated and must be followed by genetic counseling for recurrence risk.

Keywords: ultrasound, skeletal dysplasia, fetal autopsy