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Widening of the femoral proximal diaphysis–metaphysis angle in fetuses with achondroplasia

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Objective

The widening of the femoral proximal diaphysis–metaphysis angle (DMA) in achondroplasia is a relatively new sign, and there are only a few cases in the literature. Our aim was to report 3 new cases and a literature review.

Methods

Between January 2021 to April 2023, in 3 consecutive fetuses with prenatal suspicion and confirmation of achondroplasia the femur was routinely measured with the angle of insonation perpendicular to the long axis of the bone, as recommended, and the measurements were plotted on growth charts to determine the centile, using ASTRAIA. The ultrasound transducer was then positioned at an angle of 45◦ to the diaphyseal axis, and the angle between the diaphysis and the metaphysis was measured, as previously described by Boulet et al.

Results

Case 1: Dichorionic twin pregnancy, 23^{+5} weeks. Twin B showed short long bones, and the DMA was 125°, with postnatal clinical and radiologic confirmation of achondroplasia. Twin A showed normal biometry and anatomy, with a DMA of 89°, and normal postnatal evaluation. Case 2: Singleton pregnancy with normal first trimester screening and 20 week anomaly scan. At 25 weeks the scan showed rhizomelic shortening of the long bones and a DMA of 130°. Amniocentesis and molecular testing confirmed achondroplasia. Case 3: Singleton pregnancy referred with prenatal suspicion of skeletal anomaly at 31^{+6} weeks. The scan showed macrocephaly, frontal bossing, depressed nasal bridge, rhizomelic shortening of the long bones and polihidramnios. Te DMA was 125°. Literature Review: The initial description of the sign was performed by Boulet et al (Prenat Diagn 2009;29: 697–702) and included 4 cases of achondroplasia in the third trimester with DMA >130° (+3.3 SD). Khalil et al. (Ultrasound Obstet Gynecol 2014 Jul;44(1): 69-75), also in the third trimester, showed similar findings in 5/6 fetuses with achondroplasia, with a median DMA=142° (126-150°) compared to DMA in normal pregnancies -109° (103-116°) and SGA pregnancies-110° (103-116°). In 2016, Khalil et al. found at 20-23 $^{+6}$ weeks that in 4 fetuses with achondroplasia the DMA was also higher-median DMA=125° (119.8-131.8°) compared to controls-median DMA=95° (88-99°).

Conclusion

A widened femoral DMA represents a potential second and third-trimester useful and early sign for achondroplasia in fetuses with shortened long bones.