The long and short of it – antenatal clues to lethal chondrodysplasias

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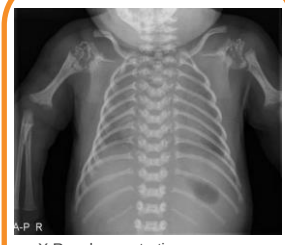
Introduction

The skeletal dysplasias are a heterogenous group of heritable bone disorders. Unfortunately, many diagnoses are not made until after birth as signs are non-specific, and most cases of short femur found antenatally represent constitutional short limb. Further differential diagnosis of short limb on ultrasound includes aneuploidy, SGA/IUGR, skeletal dysplasias, incorrect measurement and incorrect dating. Earlier identification of chondrodysplasia and lethality provides couples with an opportunity to consider options for continuing the current pregnancy, and to optimise postdelivery care. This case of rhizomelic chondrodysplasia punctate highlights several diagnostic clues to the rarer forms of lethal chondrodysplasias.



X Ray demonstrating characteristic calcific stippling of cartilage and peri-articular soft tissues seen in rhizomelic chondrodysplasia punctata. Note also shortened humerus and thoracic hypoplasia. (Image courtesy of Rick van Rijn rID40230, Radiopaedia.org)

Case

The 26yo G2P1 presented to our regional antenatal service at 26+6/40. Her combined first trimester screen was low risk, and morphology scan was normal. At 30 weeks, she was found to have an increased symphysofundal height. Ultrasound reported normal amniotic fluid with femur length below the 2nd centile. The patient was reassured, and routine pregnancy care continued. Two weeks later, polyhydramnios was again clinically suspected. Repeat ultrasound confirmed polyhydramnios and tertiary referral occurred. Other workup for polyhydramnios including TORCH serology, repeat glucose tolerance test and kleihauer were normal. Tertiary assessment confirmed short femur length with other parameters being normal (humeral length was not commented on). AFI was at the upper limit of normal at 23.2. Care was continued locally, and a diagnosis of chondrodysplasia was not made until after birth.

Discussion

Rhizomelic chondrodysplasia punctate is a form of lethal chondrodysplasia, with most children not living beyond two years. Antenatal signs of lethal dysplasias include short humeral length in addition to short femur length and a fall in femur length centiles over time. The finding of polyhydramnios in lethal skeletal dysplasias is common, postulated to be secondary to oesophageal compression due to thoracic hypoplasia. Other sonographic markers suggestive of skeletal dysplasia include femur length less than 5mm below 2SD for gestational age, femur: foot ratio of < 1, femur: abdominal circumference <0.16. This case reflects on some ultrasonographic clues that may improve antenatal diagnosis.

References

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