

## **Short long bones: prenatal ultrasound phenotype**

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### **Objective**

To detect phenotypic abnormalities by prenatal ultrasound in fetuses with short long bones.

### **Methods**

We retrospectively examined a series of 90 cases of fetuses with short long bones defined as humerus and/or femur length <5<sup>th</sup> percentile for the gestational age between 2010 and 2023. We analysed the associated ultrasound findings in each case and genetic results in the cases which underwent invasive testing. In this study, we used current growth curves to assess fetal femur & humerus length, and focused on additional fetal malformations, chromosomal defects and birth-related risks such as small for gestational age babies (SGA), preterm birth (PTB) and low birth weight (LBW) to appropriately counsel parents with affected fetuses.

### **Results**

In our study, short long bones were observed to be normal variant, marker of early onset growth restriction, soft marker for aneuploidy, indicator of skeletal dysplasia and in some cases, associated with other structural anomalies.

### **Conclusion**

This study was conducted to determine the extent to which ultrasound diagnosis of 'short long bones' is associated with poor fetal outcome. In any case of additional structural abnormality, genetic testing should be offered. If short long bones are found to be associated with small for gestational age baby, standard pregnancy care should be intensified for intrauterine growth restriction. Combining prenatal investigations with postnatal can help improve outcome of fetuses with short long bones.