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Case Report

Isolated Short Femur Length in the Third Trimester

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Femur length (FL) is one of the three key biometric parameters routinely measured via ultrasound during prenatal visits to determine fetal gestational age and growth. A short FL is generally defined as a value that is two standard deviations below the mean FL for a particular gestational age. The etiology of short FL includes skeletal dysplasias, aneuploidies, and intrauterine growth restriction. We present a case of a pregnant patient with isolated short FL of fetus on third trimester ultrasound exam.

Keywords: pregnancy; short femur length; ultrasound

Introduction

Femur length (FL) is one of the basic biometry measurements taken during prenatal sonography to assess gestational age and fetal growth. A short FL is defined as fetal FL that is 2 standard deviations below the mean FL for a particular gestational age [1]. Short FL is related to aneuploidies and skeletal dysplasia.

Case report

A 31-year-old Gravida 1 Para 0 pregnant woman received regular care at our outpatient prenatal clinic. Her and her husband's family histories were unremarkable. No significant abnormalities were noted until the 31st week of pregnancy. At that time, FL was 5.4 cm, which correlated with a gestational age of 29 weeks. Biparietal diameter (8.2 cm) and abdominal circumference (26.4 cm) were within the normal range for gestational age. Non-invasive fetal trisomy (NIFTY) results, amniotic fluid index, and placental function were all normal. Due to a lack of other significant findings, isolated short FL was suspected. Subsequent antenatal sonography showed continued isolated short FL (Table 1) and breech presentation of the fetus.

Table 1. Fetal biometric measurements
Abbreviations: GA, gestational age; BPD, biparietal diameter; AC, abdominal circumference; FL, femur length

GA	BPD	AC	FL
31+	8.2 cm (32 wks)	26.4 cm (32 wks)	5.4 cm (29 wks)
33+	8.6 cm (34 wks)	28.3 cm (34 wks)	5.2 cm (28 wks)
35+	9.0 cm (35 wks)	30.0 cm (35 wks)	5.8 cm (31 wks)
36+	9.2 cm (38 wks)	29.6 cm (35 wks)	6.2 cm (33 wks)

Due to breech presentation, the patient underwent cesarean section at 38 weeks and 5 days of gestation. A 2910g singleton male live newborn was delivered. Apgar score was 7 at the first minute of life and 8 at the fifth minute of life. Phenotypic feature of shortened crus was noted (Figure 1). Other physical features were normal. Based on the overall findings of prenatal sonography and phenotypic characteristics of the neonate, skeletal dysplasia was

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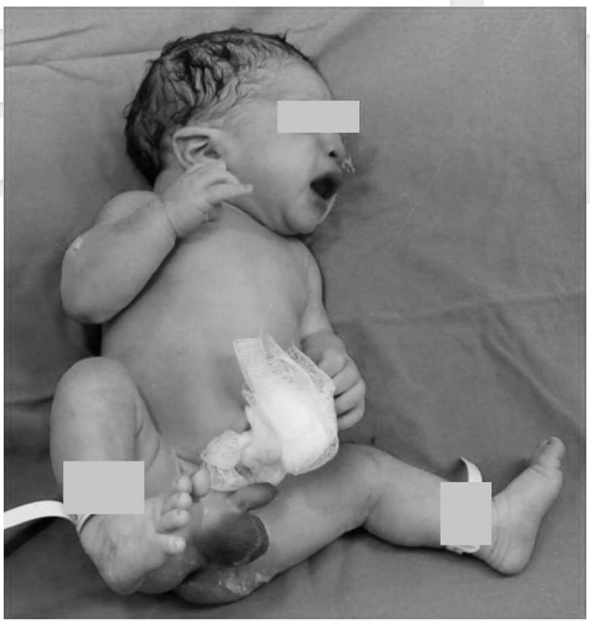

Fig. 1 Phenotypic features of the newborn

suspected. FGFR3 mutation analysis was conducted which revealed 1349 C>C/T mutation on Exon 10. Congenital metabolic disorder laboratory tests were all negative. For further analysis, paternal and maternal DNA isolation tests were sent for analysis. Mother and newborn were discharged from the hospital and regular postpartum visits were conducted at our outpatient department.

Discussion

Short femur length is generally defined as FL that is 2 standard deviations below the mean FL for a particular gestational age on ultrasound examination [1]. Past studies have shown that the prevalence of isolated short FL is about 5% [2]. Isolated short femur diagnosed in the second trimester in a fetus with previously normal findings is a cause for concern due to its relation to skeletal dysplasia (SD), various aneuploidies, and intrauterine growth restriction (IUGR) [2,3,4]. Past studies have shown that an isolated short femur significantly increases IUGR or small for gestational age (SGA) risk when diagnosed on second trimester ultrasound scans [5,6]. Table 2 lists possible diagnoses for isolated short FL and characteristics of each disorder.

When short FL is detected, a variety of causes must be investigated to develop appropriate management strategy. When compared with a high-level marker for aneuploidies, such as nuchal skin fold, short FL is a low-level marker [7]. Down syndrome typically exhibits as multiple markers of aneuploidy such as choroid plexus cysts, duodenal atresia, nuchal translucency, or pyelectasis. Past studies have shown short femur in early pregnancy in Down syndrome. Diagnosis is generally via Down syndrome screening tests and the presence of more than one soft marker on karyotype analysis during antenatal care. Since isolated short femur is considered a low-level marker, other sonographic markers are required to conduct karyotype analysis [5]. Several studies have shown an association between isolated short FL and IUGR. However, IUGR typically presents with other abnormal biometric measurements and Doppler findings.

Table 2. Causes of isolated short femur length

	Aneuploidy	Skeletal Dysplasias	IUGR
Clinical Findings	- Markers of aneuploidy (absent nasal bone, cystic hygroma, nuchal translucency, etc.)	- Tubular bones - Bowing of bones - Metaphyseal changes	- Small abdominal circumference, femur length, and/or biparietal diameter - Abnormal Doppler findings
Femur Growth	- Short femur length from early pregnancy onwards	- Short femur length at second trimester or before	- Variable onset. With or without shortened femur length
Diagnostic Tests	- Karyotype analysis if more than 1 marker is present - Aneuploidy screening tests such as NIFTY	- Assessment of all other long bones for abnormalities - Molecular testing	- Serial ultrasonography - Karyotype analysis if more than 1 marker is present

Onset of abnormal femur growth pattern in IUGR varies but generally appears with a small abdominal circumference on sonography. Diagnosis can be confirmed via serial ultrasonography to monitor growth restriction and karyotype analysis if more than one soft marker is noted during antenatal care. Skeletal dysplasia must be considered in any case of long bone shortening. There are over 450 subtypes of skeletal dysplasia, including achondroplasia and hypochondroplasia [8]. Generally, if short FL is noted on sonography, assessment of all other long bones against normal expected values is recommended. Skeletal dysplasias are normally associated with short FL before 24 weeks of gestation and accompanied by abnormalities of other tubular bones, metaphyseal changes, or bowing. Diagnosis depends on family history and molecular testing results.

In conclusion, if isolated short FL is noted on ultrasonography, a conservative management approach is recommended. Generally, isolated short FL is discovered during the second trimester. In this case, short FL was not observed until the third trimester. Serial ultrasonography to exclude skeletal dysplasia, IUGR, and aneuploidies must be conducted. If there are sonographic markers suggestive of aneuploidies, such as nuchal skin fold, karyotype analysis is recommended. Short FL due to IUGR should be suspected if sonography results reveal small abdominal circumference or abnormal Doppler parameters, especially of the uterine and umbilical arteries. Possible diagnoses, plans for the newborn, and risk factors for future pregnancies must be discussed with the parents.

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