Case report

Isolated Short Femur Length in the Third Trimester

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Abstract

A short femur length (FL) has been defined as the presence of a biometric value of fetal FL below the 5th percentile or ~2 standard deviations (SD) for gestational age at ultrasound examination. A short FL was also defined as a biparietal diameter to FL ratio above 1.5 SD for gestational age or as observed to expected FL ratio ≤0.91 for gestational age. Short FL was considered isolated when both the estimated fetal weight and abdominal circumference were >10th percentile for GA. Here, we report a case of a pregnant patient with isolated short FL of fetus in third trimester ultrasound exam.

Keywords: Pregnancy, Short femur length

Introduction

A short femur length (FL) has been defined as the presence of a biometric value of fetal FL below the 5th percentile or ~2 standard deviations (SD) for gestational age at ultrasound examination [1]. A short FL was also defined as a biparietal diameter to FL ratio above 1.5 SD for gestational age or as observed to the expected FL ratio ≤0.91 for gestational age [2, 3]. The detection of a femur length below the 5th percentile is often a diagnostic dilemma since this sonographic finding has been described as a normal variant in constitutionally small fetuses, but may also be associated with skeletal dysplasia or chromosomal abnormalities [4], especially trisomy 21. Recent studies have also suggested an increased incidence of adverse perinatal outcomes such as small for gestational age (SGA) neonates, low birth weight (LBW), and preterm birth (PTB)[4,5] because of a suspected association of short FL with placental insufficiency. While during the first trimester of pregnancy, fetal crown-rump length and biparietal diameter are used to measure and confirm gestational age, a combination of the fetal biparietal diameter, the head circumference, the abdomen diameters, and its circumference as well as the femur length of the baby is used during the second and third trimester for prenatal growth estimation and weight assessment [6]. 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. Skeletal anomalies are in the group of anomalies most difficult to be diagnosed in the antenatal period. Diagnosis of isolated femoral shortening during the second half of pregnancy has so far been considered a cause for concern as it has
been related to Down’s syndrome (DS) or Skeletal Dysplasia (SKD)[1,7,8]. If family history is not present, early diagnosis is difficult because of the late appearance of ultrasound signs of this disease. It has been suggested that an isolated short FL may be an early marker of placental dysfunction as highly oxygenated fetal blood is preferentially shunted toward vital organs such as the heart and lungs at the expense of the extremities [9]. Furthermore, a short femur is considered to be a so-called soft marker, which, in combination with other soft markers, increases the risk of trisomies, especially trisomy 21. The influence of various factors, such as maternal and paternal height, BMI or birth weight, as well as ethnicity on medical conditions, maternal demographic information, obstetrical history, maternal medical comorbidities, ultrasound findings, pregnancy complications, and neonatal outcomes were extracted from our ultrasound and genetics database of this institution (Nepal Medical College, Jorpati).

Case Report
The case is of a term male baby delivered at 38 weeks and three days of gestation to a gravida 2, parity 1 (G2P1A0L1) with no consanguinity. Regarding maternal history, the mother’s age is 35 yrs, height: 161.54 CM, and weight: 72 KG. Both the parents had achieved average height. She had regular antenatal examinations at our hospital and was immunized. Her antenatal period was uneventful. She took folic acid, iron, and calcium regularly. She has no history of any illness, smoking, and alcohol use during her pregnancy. No h/o diabetes mellitus, hypertension, thyroid disorders, consanguinity, or any congenital anomalies in the family. There was no family history of rhizomelic shortening of limbs. She has been married for 14 yrs and has a female child of 12 yrs born via normal vaginal delivery which was uneventful. At 17 weeks, QUADRUPLE TEST showed positive results for trisomy and neural tube defects whereas AMNIOCENTESIS was apparently normal. AMNIOTIC FLUID AND FISH SCREENING was done which showed normal chromosome complement with normal diploid status for chromosomes 13, 18, 21, and sex chromosomes. Anomaly scans at 13 weeks and 1 day of gestation showed no gross anomaly of the fetus. Third-trimester scan at 33+6 WOG from the last menstrual period showed a single, live, intrauterine pregnancy corresponding to 33-34 weeks 1 day of gestation with the anterior fundal placenta and cephalic presentation of the fetus with adequate liquor. The fetus had a normal growth pattern before 33 weeks of gestational age. At 33 WOG, USG was done which showed an estimated fetal weight = 1774 grams. It also noted a bilateral short femur measuring 5.0 cm corresponding to 26-27 weeks of age. At 36 WOG, USG was repeated again and femur length was seen to be 5.6cm which corresponds to 29-30 weeks of age (Table 1). Serum TORCH IgG & IgM evaluation was done which showed high titers, demonstrating an infection.

The baby was born via Elective LSCS on maternal request having an APGAR score of 8/10 and 9/10 at 1 and 5 minutes respectively and a birth weight of 2.4 kg. On examination, the baby was active, alert, and had no signs of respiratory distress. Cardiac examination revealed normal S1 and S2 with the absence of any murmur. The rest of the system did not give any other abnormal findings. Head to Toe examination: no abnormality seen. USG and ECHO findings were found to be normal. XRAY (infantogram) was done on the 7th day of life, which showed slightly shortened bilateral femur measuring 57mm (corresponding to 33 WOG), epiphyseal region appears normal. His vitals were stable and reflexes were preserved. Mother and newborn were discharged from the hospital and regular postpartum visits were conducted at our outpatient department and were asked to follow up with the reports and further evaluation. As serum TORCH findings showed high IgG titers, an ophthalmic evaluation was done which showed unremarkable findings.

Table 1: Table showing the gestational age, BPD, AC, and FL of the fetus with the corresponding gestational age, according to the weeks of gestation

<table>
<thead>
<tr>
<th>Gestational Age (GA)</th>
<th>Biparietal Diameter (BPD)</th>
<th>Abdominal Circumference (AC)</th>
<th>Femur Length (FL) and corresponding gestational age</th>
</tr>
</thead>
<tbody>
<tr>
<td>13+</td>
<td>2.6 cm (13 wks)</td>
<td>7.0 cm (13 wks)</td>
<td>1.0 cm (13 wks)</td>
</tr>
<tr>
<td>18+</td>
<td>4.1 cm (17 wks)</td>
<td>12.1 cm (17 wks)</td>
<td>2.5 cm (17 wks)</td>
</tr>
<tr>
<td>33+</td>
<td>8.3 cm (33 wks)</td>
<td>29.1 cm (33 wks)</td>
<td>5.0 cm (26-27 wks)</td>
</tr>
<tr>
<td>36+</td>
<td>9.0 cm (36 wks)</td>
<td>32.0 cm (36 wks)</td>
<td>5.6 cm (29-30 wks)</td>
</tr>
</tbody>
</table>

Discussion
This case report was aimed to evaluate the strength of association between isolated short femur length (FL) and fetal growth restriction (FGR) and other adverse perinatal outcomes. The hypothesis that an isolated short FL might be considered an early marker for placental dysfunction was taken to note. One potential mechanism is that abnormal placentation leads to
altered secretion of fibroblast growth factor 2 (FGF-2). FGF-2 is normally secreted by the human placenta and plays a role in skeletal development. Altered secretion of this growth factor could result in inhibition of long bone growth in the fetus [10, 12]. Another potential mechanism involves the fetal adaptive response to chronic hypoxia in which highly oxygenated blood is shunted toward vital fetal organs such as the brain and heart at the expense of the extremities [9]. It should be underlined that in the case of isolated short femur, conservative counseling should be given to the parents because an uncomplicated pregnancy with a normal neonate at term has been reported in majority of cases [13]. Obstetricians should use a more careful approach to the management of these fetuses in order to prevent perinatal adverse outcomes, monitoring these pregnancies in a tertiary center [9, 13, 14, 15, 16] with a structured program of sonographic surveillance and accurate delivery [17]; when malformations are excluded, serial ultrasound studies of fetal growth and frequent monitoring of maternal blood pressure are indicated. 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 [11]. Skeletal dysplasia must be considered in any case of long bone shortening. There are over 450 subtypes of skeletal dysplasia, including achondroplasia and hypochondroplasia [11]. 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. Generally, if short FL is noted on sonography, assessment of all other long bones against normal expected values is recommended. Diagnosis depends on family history and molecular testing results.

Conclusion
The finding of a short FL during ultrasonography should be followed by further Ultrasound Scan to exclude fetal malformations. Typically, such occurrences are identified during the second trimester, but in this instance, the short FL was not observed until the third trimester. If there are ultrasound indicators that suggest aneuploidies, such as nuchal skin fold, it is recommended to conduct karyotype analysis. In cases where a short femur is part of a small-for-gestational-age baby, intrauterine growth retardation, or suspected late growth retardation, standard prenatal care should be augmented with additional interval biometry and Doppler assessment every two weeks. In this report, we present one of the rare cases of the fetus with short femur length in the third trimester.

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Conflict of interest: None

References
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