PRENATAL ULTRASONOGRAPHIC DIAGNOSIS OF BILATERAL PROXIMAL FEMORAL FOCAL DEFICIENCY WITH CLEFT LIP AND CLEFT PALATE: FEMORAL HYPOPLASIA/UNUSUAL FACIAL SYNDROME

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ABSTRACT

Antenatal ultrasound has pivotal role in early and accurate assessment of skeletal and nonskeletal congenital anomalies of fetus that help in future management and prognosis of pregnancy outcome. Proximal femoral focal deficiency (PFFD) represents a rare and complex deformity manifested by hypoplasia of a variable portion of the femur with shortening of the entire limb. The condition may be unilateral or bilateral and is often associated with other congenital anomalies. We report a case of fetal non-familial PFFD along with facial abnormalities diagnosed prenatally using two-dimensional images.

KEYWORDS

Congenital; Bilateral Femoral Deficiency; Foetus; Ultrasound, Femoral Length, Cleft Lip and Cleft Palate.


CASE REPORT

A 28-year-old woman, non-diabetic primigravida was referred to ultrasound department for antenatal routine sonography at 19 weeks' gestation.

Detailed 2D Ultrasonography was done using a Xario (Toshiba, Japan) sonography machine using 3.5 Mhz convex array probe. Gray-scale 2D examination showed a markedly asymmetrical shortening of the bilateral femur, which measured 13.0 mm in length (Consistent with a gestational age of 13 weeks 6 days).

Marked bowing of femoral shafts was noted. These values were below 5th percentile for average gestational age. Rest of long bones and feet appear unremarkable and shows measurements consistent with average gestational age. All Bones showed normal echogenicity. Tibia and ulna measured 28 and 24 mm in length respectively (Consistent with a gestational age of 19 to 20 weeks).

Biparietal diameter and abdominal circumference, rest of bones were all consistent with menstrual dates.

Detailed evaluation of face revealed bilateral cleft lip with cleft palate.

Detailed clinical history of patient showed non-consanguineous marriage no significant relevant family history or prior history of miscarriage. No prior history of exposure to radiation or teratogenic drugs was noted. Hence the etiology was unknown in our case.

Patient was counseled regarding fetal outcome and referred to obstetrics and gynecology department for further management.

PFDD, which occurs in 1–2 per. 100000 births.1,2 is a congenital developmental disorder of the subtrochanteric region of the femur with varying degrees of shortening of the proximal femoral segment that causes hip deformity and altered function of the involved lower extremity. It may be bilateral in 15% of cases.

Congenital hypoplasia of the femur is a principal sign of 4 uncommon malformations: (a) PFFD, (b) femur/fibula/unlar hypoplasia (FFU), (c) femoral hypoplasia/unusual facial syndrome (FH/UFS), and (d) limb/pelvis-hypoplasia/asplia syndrome.

Musculoskeletal conditions that may be observed in this condition are absent fibula (50–80%), short tibia and fibula, patellar abnormalities, limb malrotation, and proximal joint instability and absence of anterior and posterior cruciate ligaments. In up to 70% cases other systemic anomalies are detected, such as cleft palate, clubfoot, congenital heart defects, and spinal anomalies.

The etiology of PFFD is uncertain, but it may be due to a developmental insult at the time of limb bud formation at 4–6 weeks’ gestation.3

Additional etiologic factors include poor diabetic control, exposure to drugs (Thalidomide), viral infections, radiation, focal ischemia and trauma between the 4th and 8th weeks of gestation.4,5

Prenatal sonographic diagnosis of PFFD has been reported in the literature, and was described for the first time in 1989.6 Since then other authors have described prenatal detection of bilateral and unilateral proximal femoral hypoplasia with 2D and 3D ultrasound 4 and 3D images produced by computed tomography7 between 18 and 32 weeks’ gestation. MRI also has been also used in classifying PFFD.8

Management of PFFD requires a multidisciplinary and highly individualized approach based on leg-length discrepancy, adequacy of proximal musculature, femoral rotation and proximal joint stability and ranges from limb salvage, lengthening and hip reconstruction in mild cases to amputation and prosthetic fitting in severe cases.9,10
Hence for early detection of this condition careful assessment of bilateral femur length is important. When length shortening is observed in femora, other bones should be carefully assessed. A thorough assessment of face, thorax and spine helps in detection of systemic abnormalities.

**Fig. a:** En profile view shows bilateral paramedian defect in upper lips and hard palate

**Fig. b,c:** Marked shortening of bilateral femora and lower limbs

**Fig. d:** Marked shortening of bilateral femur length as compared to normal tibial length

**CONCLUSION**

In conclusion, bilateral PFFD with facial abnormality is rare condition and is part of femoral hypoplasia/unusual facial syndrome. In this case 2D ultrasound was instrumental in providing accurate pre-natal evaluation. Early recognition and exact pre-natal diagnosis of this condition may provide the prospective parents with a proper evaluation of their fetus and will ensure the acquisition of useful information for a therapeutic plan.

**REFERENCES**