



Prenatal Ultrasonographic Diagnosis of Proximal Focal Femoral Deficiency

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Abstract

This is a methodological type research that was proceeded to evaluate the validity and reliability of the Scale for the Problematic Mobile Phone Use (PU) among the Turkish society. The related research was conducted within the Spring Semester of 2011-2012 Academic Year on 387 students attending Inonu University Medical Faculty. Besides the scale, a questionnaire for determining the descriptive features of the students was also used in the study. The German original scale was translated into Turkish by using the group translation and retranslation techniques. Content validity index were analyzed for the content validity, exploratory and confirmatory factor analyzes were conducted for the construct validity and of the scale, the stability over time and internal consistency for reliability analyzes were conducted. Turkish questionnaire of the scale was reviewed by 10 experts to determine for the content (theme) validity. The necessary corrections were done in accordance with the suggestions of the experts, and the value of the content validity index of the scale (KGI) was found 0.89. Exploratory and confirmatory factor analyses performed to test the structure validity of the scale were completed, and a three-factor structure explaining the 45% of the total scale variance was obtained. Cronbach Alpha value calculated for the reliability analyses of the scales was found 0.854, meanwhile the test-retest correlation coefficient of the scale for the total points was found 0.86. Additionally, the difference between the total average points of the pre-test and the re-test of the scale was not found statistically significant ($p=0.30$). As a result of the Turkish version of the scale of the values obtained from the PU is defined to be a reliable and valid scale.

Key Words: Femoral, hypoplasia, ultrasonography, anomalies, prenatal

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Introduction

Proximal focal femoral deficiency (PFFD) is a condition where the femur is short and associated with an apparent loss of continuity between the shaft and neck. The condition may be unilateral or bilateral and is often associated with other congenital anomalies. It is important to determine whether isolated femoral abnormality is part of the syndrome. Recent technological advances in ultrasound imaging offer the opportunity to detect an increasing number of rare skeletal malformation syndromes whose correct diagnosis is essential for adequate counselling and management of the pregnancy. Incidence of PFFD is low and unilateral femoral hypoplasia has an incidence of 1.1–2 in 100 000 live births [1].

Case report

23 yrs old primigravida at 19+3 weeks period of gestation (POG) reported to fetal medicine out patients department (OPD) with an ultrasound report showing unilateral short femur. This was her first pregnancy and she gives no history of fever in last six months. The parents were nonconsanguineous and there was no family history of skeletal dysplasia. Double marker test result revealed a low risk for trisomy and diabetes screening was normal. Ultrasonography showed a markedly asymmetrical shortening of the right femur, which measured 18.7 mm in length (consistent with a gestational age of 14+3 weeks) and a normal left femur measuring 32.1 mm in length (consistent with a gestational age of 20 + 0 weeks) (Figure 1). There was also absent fibula on the right side. No other skeletal abnormalities; thorax anomalies; or cardiac, gastrointestinal, genitourinary, and neurological signs of fetal abnormalities were observed. On the basis of the ultrasound findings PFFD was felt to be the probable diagnosis. After parental counselling, the couple decided to terminate the pregnancy. Mid trimester abortion was carried out by misoprostol application and examination of the abortus confirmed the ultrasonographic findings (Figures 2 and 3).

Discussion

Proximal femoral focal deficiency (PFFD) is a developmental defect of the proximal femur and acetabulum. It includes a spectrum of findings ranging from mild femoral shortening with varus deformity to complete absence of both acetabulum and proximal femur with only a small distal

fragment [2]. The unilateral form is approximately 85–90% of all cases and although a few familial cases have been described but mostly it is sporadic and its genetic transmission mode is unknown [3]. It is important to examine the face carefully because the disorder may be bilateral femoral hypoplasia and unusual facies syndrome in cases where both the femora are affected [4] which consists of bilateral femoral hypoplasia and facial anomalies including short nose with broad tip, long philtrum, microgathia and cleft palate. Long bone abnormalities can extend to other segments of the upper and lower extremities like absent fibula which was seen in our case. As 4-8 weeks period of gestation (POG) is critical for skeletal development, poor diabetic control in the early weeks of pregnancy, drug exposure (thalidomide), viral infections, radiation, focal ischemia, chemical toxicity, trauma, and causes of familial transmission are some of the etiologic factors [5].



Figure 1. Ultrasonography shows different size of femurs



Figure 2. External photograph of femurs

Except in bilateral proximal focal deficiency, the diagnosis is usually suspected when there is unilateral short femur, in the absence of other anomalies. Prenatal diagnosis of syndromes associated with abnormalities of the femur is possible; however, although only 19% of cases have been diagnosed prenatally, 68% of cases have been diagnosed postnatally [6]. When a short femur is detected, a differential diagnosis should be made with other skeletal dysplasias. Ultrasonographic diagnosis of femoral hypoplasia is possible after the second trimester of

pregnancy. Some cases diagnosed at approximately 14 weeks of gestation by transvaginal ultrasonography have been reported in the literature [7]. During ultrasonographic examination, length discrepancies and disproportion between femurs and other bones are the diagnostic determiners of PFFD. The Aitken classification, which is the most widely used, divides PFFD into four categories based on the radiographic appearance ranking from a benign form (A) to a severe form (D) according to the presence of the femoral head, a stable hip joint, or acetabular hypoplasia [8]. This classification does not take into account the classification of cartilage and soft tissue abnormalities.



Figure 3. X ray of abortus showing the size difference of femurs

According to Paediatric Orthopaedic Society of North America (POSNA), treatment of PFFD requires complex decision-making, surgical reconstruction, and prosthetic expertise. Milder forms can be treated with correction of the proximal pseudarthrosis, valgus osteotomy of the proximal femur, and femoral lengthening. In more severe cases, amputation and prosthetic fitting is necessary. There are several surgical approaches currently employed to facilitate prosthetic fitting and improve function. Selection of procedure and timing are critical to optimizing function of patients with PFFD. Fitting with a prosthesis modified to accommodate the foot and leg is often prescribed at about 12 months, when the child would developmentally start ambulating [9, 10]. Children in developing countries like India who have limited access to medical care will generally find a way to ambulate and participate in childhood play. Beginning care later in childhood is

much more difficult. Genetic counselling for children with limb deficiencies is also difficult, as predictable genotype-phenotype relationships have not yet been established.

In conclusion, PFFD is rare and early recognition and exclusion of skeletal dysplasia should be aimed during anomaly scan at 18- 20 weeks POG as exact prenatal diagnosis of this condition will help in counselling which may provide the prospective parents with a proper evaluation of their fetus and will ensure the acquisition of useful information for a therapeutic plan.

Conflict of interest: None identified

Consent: Written informed consent was taken from the patient for publication of case report.

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