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Congenital Deficiency of The Proximal Femur
Literature Review with a Case Report

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✔ This is a case of a 20-year-old female patient with unilateral proximal focal femoral deficiency. Our case report is important since the patient had a normal tibia and fibula. The patient's mother had no gestational history of any drug ingestion, infection, diabetes mellitus or other conditions that could be associated with the malformation. There were no other cases in the family. The patient's parents were not consanguineous. Chromosome analysis was normal.
This review comprises the literature related to partial or complete deficiency of the femur from 1959 to 1998.
Key words: *Femur, congenital deficiency, hypoplasia*

✔ **Femur Üst Ucunun Doğumsal Gelişim Yetersizliği: Bir Vaka Sunumu ve Kaynakların Gözden Geçirilmesi**
Makalemizde femur üst ucunda tek taraflı gelişim yetersizliği olan 20 yaşındaki bayan hasta sunulmaktadır. Hastanın tibia ve fibula'sı normal yapıda olduğundan bu vakamız önemlidir. Hastanın annesinin gebelik döneminde herhangi bir ilaç kullanımı hikayesi, enfeksiyon, şeker hastalığı (diabetus mellitus) veya gelişim bozukluğu yapacak herhangi bir sebep yoktu. Ailede buna benzer başka bir vaka belirlenemedi. Hastanın anne babası arasında akraba evliliği söz konusu değildi. Kromozom analizi normaldi.
Ayrıca derlememiz 1959'den 1998'e kadar olan kaynaklarda parsiyel veya komplet tip femur gelişim yetersizliği ile ilgili olarak yayınlanan çalışmaları özetlemektedir.
Anahtar kelimeler: *Femur, femur doğumsal gelişim bozukluğu, femur hipoplazisi, kısa femur*

INTRODUCTION

Congenital deficiency of the femur (CDF) includes a wide spectrum of developmental anomalies, including congenital short femur, coxa vara, and partial to complete absence of the femur, also known as proximal focal femoral deficiency (PFFD). Patients with CDF have traditionally been examined clinically and radiographically to determine whether osseous and soft tissue elements are present, then treated accordingly[1-3]. In young infants with femoral deficiency, ossification is often delayed or abnormal which limits radiographic evaluation[3].

According to Goldman et al., proximal focal femoral deficiency (PFFD) is applied to a

spectrum of conditions characterized by partial absence and shortening of the proximal femur and thought to result from early disturbance of growing mesenchyme[4]. Most cases of femoral hypoplasia, also known as proximal focal femoral deficiency or congenital deficiency of the femur are unilateral and associated with other defects of the extremities[5-8]. This situation is usually sporadic[7,9] however, familial cases have been described[5,10]. Maternal diabetes [9,11] and use of thalidomide[6] may also be related to the condition. Congenital short femur (CDF) is a rare and confusing defect ranging from simple hypoplasia to total absence of the femur[6]. A case with the combination of left proximal focal femoral deficiency, left fibula dimelia, polydactyly and congenital heart disease was reported in the literature[12]. Embryological development of these cases could not be defined.

The CDF (or PFFD) may be unilateral or bilateral. CDF is often associated with other congenital anomalies. The incidence of this complex defect was reported by Rogola et al., from Edinburg Register of the Newborn as one per 52029 of the population (0.002%)[6,13,14].

CASE REPORT

A 20-year-old girl was first seen in our clinic on 1996. She was the third child of normal parents and other children were normal. There was no history of drug ingestion, prenatal illness, or abnormality of pregnancy.

We examined all extremities of the patient. The right lower extremity and both upper extremities were normal. When she was born, there was an 8-cm-difference between her two lower extremities. When she appointed for the examination, she was 20 years old. Her right and left femur were measured as approximately 36 and 13 cm, respectively. That is 23 cm of length difference was detected between her two lower extremities. Tibia and fibula were normal and functional. On the left side knee joint was functioning normally, but hip joint had a flexion contracture of 5 degrees.

Radiological findings indicated deficiency of proximal femur on the left side (Fig. 1). There was no normal head of left femur and no regular articulation between the head of femur and acetabulum.

On examination, she appeared alert with normal mental development and no history of visceral or internal organ abnormality.

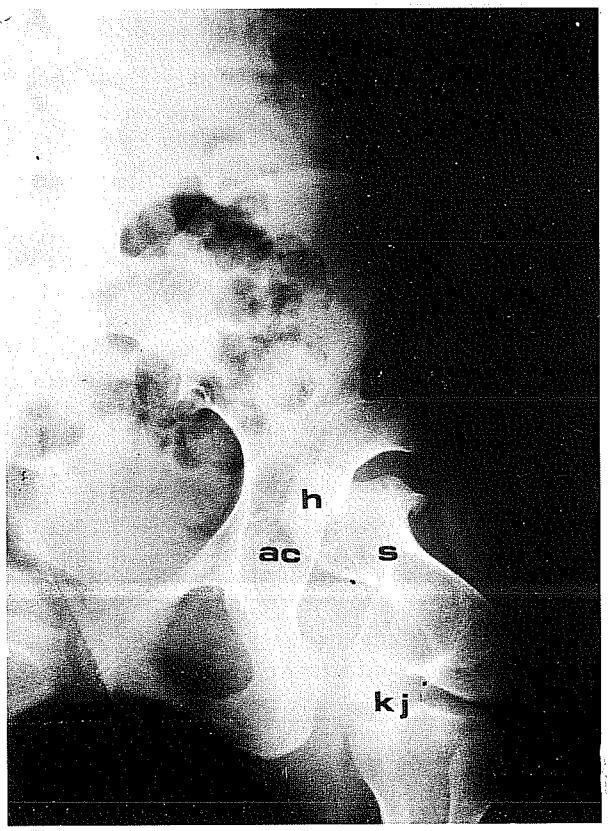

Figure 1. Abnormal head and shaft of femur
ac: acetabulum,
h: head of femur
s: shaft of femur
kj: knee joint

RESULTS

CDF is rare and many classification systems have been developed over the years. Different techniques have been used for treatment. If the hip joint is stable, the gait will be much better. Diagnosis is made with MRI, CT scan, ultrasound, etc. and early diagnosis is important for treatment. Alternative ways of treatment must be explained to the family, being more effective in early ages. Finally, the treatment team should include an orthopedist, physiotherapist and psychiatrist.

Even 1 mm difference in the lenght of right and left femur can be measured by a typical modern machine[20].

DISCUSSION

Congenital anatomic abnormalities of the femur has been reported and reviewed by different investigators. The intact femur is approximately 60% of the length of the normal leg[18]. In addition, bifurcation of the distal femur with or without tibial or fibular malformations has been reported[21,22,23].

On the other hand many authors have stated that bifurcation of the distal femur was always associated with a malformation or aplasia of one of the bones of the lower leg [21,22,23].

According to Wolfgang, the first description was by Otto in 1841, more than fifty years before the discovery of roentgenograms. Associated anomalies are common and have included ipsilateral bifurcation of the left femur[23]. According to Ostrum et al. the first case of bilateral femoral bifurcation was reported by Erlich in 1885[22]. According to Rogala et al., in modern orthopedic literature, incomplete duplication of both femur and double acetabulum was reported in 1931 by Nitche[14]. Fibular hemimelia has been reported in association with total duplication of the femur, but never with the bifurcation of the femur[24]. Burkus and Ogden dissected 103 prenatal femur and found one femur from a fetus of 7 weeks of gestation with two centers of primary ossification[22]. In 1959, Ring reported one pair of concordant twins, both of whom had hypoplasia of the femur[24]. In 1960, the Salzer reviewed 181 cases of tibial hemimelia from the world literature and reported 3.3% congenital dislocation of the hip and 1.6% proximal focal femoral defect[21,23]. There is often deficient or abnormal ossification of the proximal femur in femoral anomalies, as well as increased incidence of associated developmental dysplasia of the hip[3].

In our case, the left femur was 23 cm shorter than the right one and only femoral condyles were intact on the left side. Tibia and fibula were normal, knee joint was functional, but hip joint had contraction with 5 degrees flexion on the left. Abduction-adduction, flexion-extension and supination-pronation were restricted in the left hip joint when compared to the right.

It was postulated that congenital anatomic anomalies of the lower leg such as femoral bifurcation and tibial hemimelia resulted from lack of innervation from the L4, L5 spinal nerves. On the contrary, Ostrum et al. stated that their case did not substantiate the theory that bifurcation of the distal femur was always associated with distal anomalies secondary to lack of normal proximal-distal differentiation[21,22].

In 1974, Kelly described familial occurrence in one-third degree relative, the index patient having a curved femur[25].

The term FFU (femur-fibula-ulna) complex has been proposed for cases in which the femur, fibula and/or ulna show defects, which tend to be associated. These cases are usually sporadic. There is a marked asymmetry in the presence and degree.

Malformations are more often unilateral than bilateral, and upper limbs are affected more often than lower ones[26].

Many classification systems have been developed over the years. The best known classifications are those of Ring, 1959[24]; Aitken, 1969[1]; Amstutz, 1969[2]; Fixen and Lloyd, 1974[15]; Kostuik et al., 1975[16]; Hamanishi, 1980[6] (Figure 2); Pappas, 1983[17]; Gillespie and Torede 1983[18]; Kalamchi et al., 1985[19]; and Grissom and Harcke 1994[3] (Figure 3).

The type of femur in our case is classified as Type B by Aitken, as Type IVg by Hamanishi, as Class V by Pappas, as Type 2 by Fixen and Lloyd (Figure 4).

The principle of rotating the foot was first used by Borggreve in 1930, in a patient with a short femur secondary to infection[16].

The patients with dysgenesis of the proximal part of the femur must be differentiated from those in whom coxa vara is the manifestation of a generalized growth disturbance, such as dysplasia epiphysialis multiplex, achondroplasia, and osteochondrodystrophy, since the natural course of the deformity is usually much more benign in these conditions[27].

Recently, different techniques are used for treatment. These include; Van Nes Rotation-plasty, Rotation-Plasty of the lower limb through the knee with Simultaneous Knee Fusion Technique of Torode Gillespie, Knee Fusion for Prosthetic Conversion Technique of King in proximal focal femoral deficiency, Syme's Amputation Technique [28].

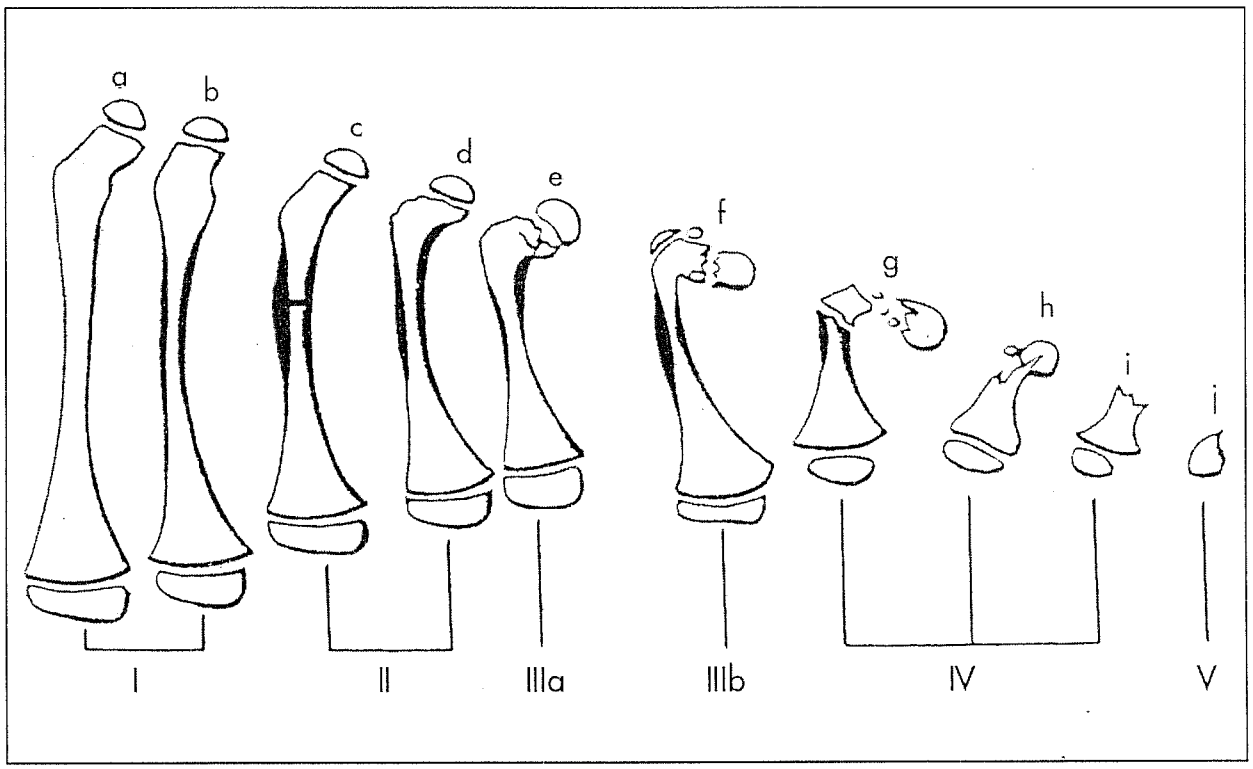

Figure 2. Classication of congenital short femur from Hamanishi (Hamanishi C. Congenital short femur. The Journal of Bone and Joint Surgery, 1980 Aug; Vol. 62-b, No. 3: 307-320) Reproduced with author's permission

Van Nes rotational osteotomy was not performed in patients with bilateral proximal focal femoral deficiency[16].

Van Nes procedure should enable the child with Syme's amputation and congenital short femur or proximal focal femoral deficiency who does not have a foot upon which he can walk, to walk without a prosthesis, as well as it provide active knee joint motion in a normal fashion[16].

Kostuik et al. believe that Van Nes procedure should not be performed to the patients under twelve years old. When it is conducted earlier, rerotation of the foot during the rapid growth period will be inevitable. According to them, younger children should be treated with an extension prosthesis until their extremities have a chance to adapt to the natural growth rate. A Syme amputation can still be performed to save the condition[16].

The treatment of isolated unilateral proximal focal femoral deficiency with rotationplasty resulted in a more energy-efficient gait than Syme amputation, with no difference in perceived physical appearance or gross function[29].

The advantages of leg lengthening procedures in younger children are early

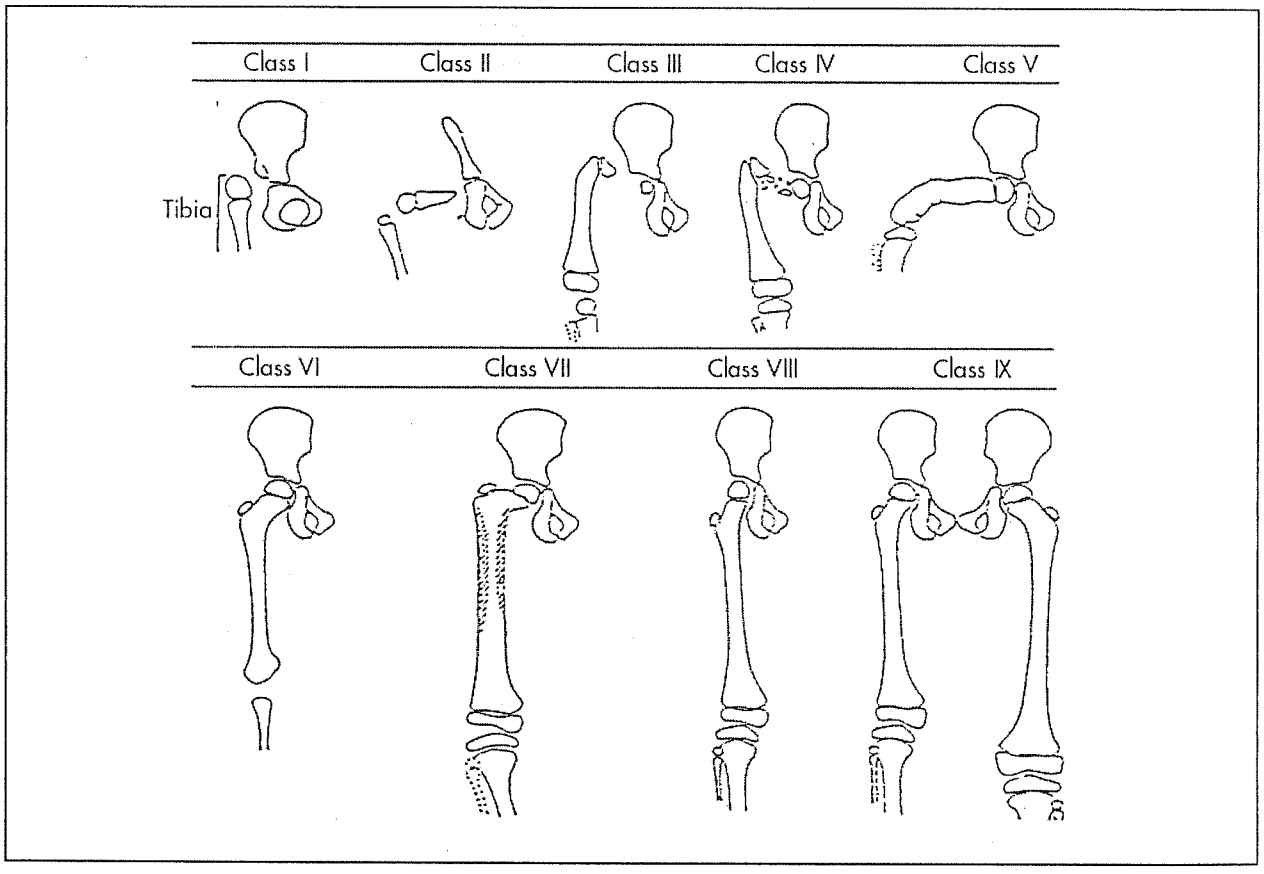

Figure 3. Classication of congenital short femur from Pappas (Pappas AM. Congenital abnormalities of the femur and related lower extremity malformations. The Journal of Pediatrics Orthopedics, 1983 Feb; 3 (1): 45-60) Reproduced with author's permission.

determination and technical ease of the application[30].

Recently the use of CT scans and MRI in the follow up of patients have proved to be of great value.

According to Kostuik et al., since the extent of the cartilaginous anlage in patients with a severe degree of focal femoral deficiency cannot be determined, authors believe that younger children should be treated with an extension prosthesis until their limb had a change to declare itself [16].

Our case report is important since the patient had a normal tibia and fibula. To our way of thinking clinicians should make a series of knee prosthesis and operations regarding this case, and make some attempts planing what to do.

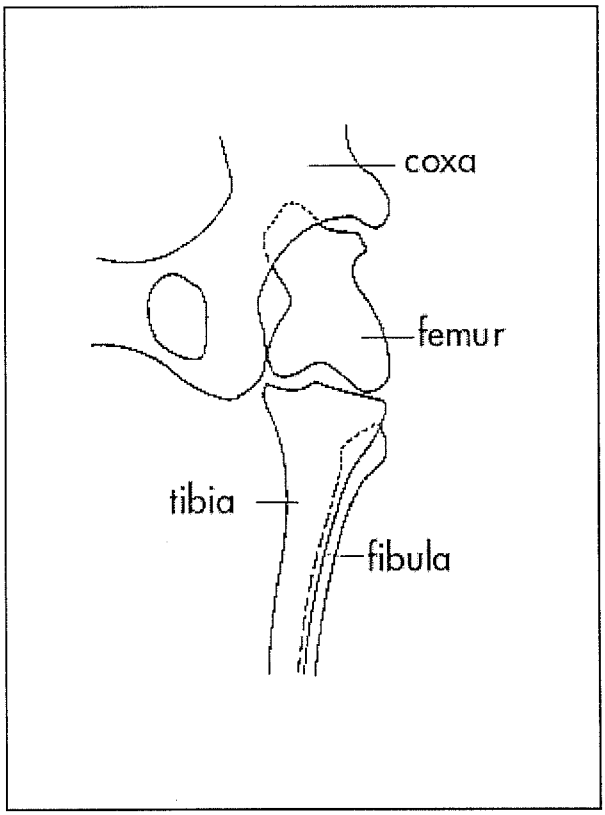

Figure 4. Schematic representation of Figure1.

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REFERENCES

1. Aitken GT. Proximal femoral focal deficiency and definition classification and management. In: Aitken, GT ed., Proximal femoral focal deficiency. A congenital anomaly. Washington, DC National Academy of Sciences 1969; 1–22.
2. Amstutz HC. The morphology, natural history and treatment of proximal femoral focal deficiency. In: Aitken, GT ed. Proximal femoral focal deficiency. A congenital anomaly. Washington, DC National academy of Sciences 1969; 50–76.
3. Grissom LE , Harcke HT. Sonography in congenital deficiency of the femur. J Pediatr Orthop 1994; 14: 29–33.
4. Goldman AB, Schneider R, Wilson PD. Proximal focal femoral deficiency. J Can Assoc Radiol 1978; 29: 101–107.
5. Connor JM, Rae PS, Connor RAC. Monozygotic twins concordent for congenital short femur. Am J Med Genet 1986; 23: 363–368.
6. Hamanishi C. Congenital short femur; clinical, genetic and epidemiological comparison of the naturally occuring condition with that caused by Thalidomide. J Bone Joint Surg Br 1980; 62: 307–320.
7. Hilman JS, Mesgarzadeh M, Reversz G, et al. Proximal femoral focal deficiency: Radiologic analysis of 49 cases. Radiology 1987; 165: 769–773.
8. Jeantly P, Kleinman G. Proximal femoral focal deficiency. J Ultrasound Med 1989; 8: 639–642.
9. Bailey PA, Beighton P. Bilateral femoral dysgenesis. Clin Pediatr 1970; 9: 668–674.
10. Gupta DKS, Gupta SK. Familial bilateral proximal femoral focal deficiency. Report of a kindred. J Bone Joint Surg 1984; 66: 1470–1472.

11. Hitti IF, Glasberg SS, Jones DH et al. Bilateral femoral hypoplasia and maternal diabetes mellitus. Case report and Review of literatures. The Pediatr Pathol 1994; 14: 567–574.
12. Igou RA, Kruger LM. Fibula bimelia in association with ipsilateral proximal focal femoral deficiency and polydactyly. Clin Orthop 1990; 258: 237–241.
13. Grill F, Dungl P. Lengthening for congenital short femur. Results of different methods. J Bone Joint Surg Br 1991; 73: 439–447.
14. Rogala EJ, Wynne DR, Littlechn A, et al. Congenital limb anomalies; frequency and aetiological factor. Data from the Edinburg Register of the Newborn. J Med Genet 1974; 11: 221–233.
15. Fixen JA, Lloyd RGC. The natural history and early treatment of proximal femoral dysplasia. Br J Bone Joint Surg 1974; 56 B: 86–95.
16. Kostuik JP, Gillespie R, Hall JE, et al. Van Nes rotational osteotomy for treatment of proximal femoral focal deficiency and congenital short femur. Am J Bone Joint Surg 1975; 57: 1039–1046.
17. Pappas AM. Congenital abnormalities of the femur and related lower extremity malformations. J Pediatr Orthop 1983; 3: 224–226.
18. Gillespie R, Torode IP. Classification and management of congenital abnormalities of the femur. J Bone Joint Surg 1983; 65B: 557–568.
19. Kalamchi A, Cowell HR, Kim KI. Congenital deficiency of the femur, J Pediatr Orthop 1985; 5: 129–134.
20. Jago JR, Whittingam TA, Heslop R. The influence of ultrasound scanner beam width on femur length measurements. Ultrasound in Med and Biol 1994; 20: 669–703.
21. Kotakemori K, Ito J. Femoral bifurcation with tibial aplasia. Clin Orthop 1978; 135: 26–
22. Ostrum RF, Betz RR, Clancy M, et al. Bifurcated femur with a normal tibia and fibula. J Pediatr Orthop 1987; 7: 224–226.
23. Wolfgang GL. Complex congenital anomalies of the lower extremities; femoral bifurcation, tibial hemimelia and diastasis of the ankle. Am J Bone Joint Surg 1984; 66-A: 435–458.
24. Ring PA. Congenital short femur; simple femoral hypoplasia, Br J Bone Joint Surg 1959; 41: 73–75.
25. Keely TE. A Familial proximal focal femoral deficiency of the femur, J Pediatr Orthop 1985; 5: 129–134.
26. Lenz W, Zygulska M, Horst J. FFU complex: an analysis of 491 cases. Hum Genet 1993; 91: 347–356.
27. Amstutz HC, Wilson PD. Disgenesis of the proximal femur (coxa vara) and its surgical management, Am J Bone Joint Surg 1962; 44: 1–21.
28. Tachdjian MO. Pediatric Orthopedics. Volume 1, Second Edition, Philadelphia WB Saunders Company, 1990: 570-580, 630–637.
29. Alman BA, Krajbich JI, Hubbard S. Proximal femoral focal deficiency. Result of rotationplasty and syme amputation. Am J Bone Joint Surg 1995; 77: 1876–1882.
30. Hope PG, Crawfurd EJP, Catteral A. Bone graft following lengthening for congenital shortening of the lower limb. J Pediatr Orthop 1994; 14: 339–342.