- Summary -

Proximal femoral focal deficiency (PFFD) is a spectrum of abnormalities ranging from minimal hypoplasia of the femur to near total absence of the femur with variable deficiency of the ilio-femoral (hip) joint. The deficiencies may include inequality of femoral lengths, malrotation of the proximal femur, instability of the hip joint, and inadequacy of the proximal hip and thigh musculature .

PFFD may be associated with other congenital skeletal anomalies . The most frequently associated congenital anomaly is longitudinal deficiency of the fibula ***(Grogan et al., 1987)*** .

There are several published classifications of PFFD . All of these classification schemes are dependent on radiographic appearance, which may be difficult to appreciate in the inmature skeleton .

The **Aitken classification (1969)** divides PFFD into four groups based on radiographic appearance ***(Aitken, 1969)*** .

Gillespie and **Torode (1983)** divided PFFD into two groups on the basis of whether the hip and knee could be functional at maturity ***(Gillespie & Torode, 1983)*** .

Pappas (1983) developed a complex classification scheme that divided PFFD into nine different classes on the basis of specific femoral and pelvic abnormalities ***(Pappas, 1983)*** .

When early radiographs have been difficult to interpret because of delayed ossification, magnetic resonance imaging (MRI) can be used to demonstrate the presence of tissue not visualized on plain radiographs ***(Grogan, et al., 1987)*** .

Typically, clinical presentation is one of a shortened femoral segment that is more bulky than the normal side, with external rotation deformity .

In more than 15% of cases, some associated anomalies of the lower leg exist, such as fibular hemimelia and shortened tibia . Often there is anteroposterior instability of the knee joint because of absence of the anterior cruciate ligament . Frequently, a valgus deformity of knee joint exists . If fibular deficiency exists, it may be associated with a ball and socket ankle joint and fusion of some of the tarsal bones . The cause for the external rotation of the whole extremity is retroversion of the femoral neck and of the acetabulum .

These associated anomalies-especially the pathomorphology and instability of the knee and hip joint - are important considerations in planning the treatment for these children in order to lengthen a congenital short femur ***(Grill, Dungl, Steinwender and Hosny, 1993)*** .

Treatment of a unilateral deformity may be nonoperative, using a nonstandard extension prosthesis that takes into account the flexed, shortened thigh and knee . The patient and his or her parents must understand that this type of prosthesis is not as cosmetically pleasing as a standard above-knee prosthesis .

According to **Epps**, the surgical treatment of PFFD must be individualized and based on several fundamental decisions .

1) Whether the patient will benefit from surgery ;
2) Whether surgery is needed to enhance hip stability (as in Aitken types A and B) ;
3) If amputation is necessary to promote function, and which type of residual limb will conversion surgery produce .

If a below-knee level is chosen, then the Van Nes rotationplasty is performed . If the above-knee level is chosen, a Syme amputation can be performed along with knee fusion and possible epiphysiodesis of the knee .

In Aitken types A and B, a procedure to increase hip stability may be indicated . This effort to improve hip stability must be tempered with possible decreased hip range of motion and the observation that patients who have not had surgical intervention have painless hips .

Operative treatment of a severe unilateral deformity usually involves ablation of the foot with a Syme-type amputation . A Boyd-type amputation is another option and is preferred by some authors because it preserves the calcaneus, which is then fused to the tibia . The somewhat bulbous stump of the Boyd-type amputee may aid in suspension of the prosthesis .

Limb length equalization may be achieved in unilateral PFFD by partial excision of the epiphyses at the time of knee fusion, epiphysiodesis, ipsilateral lengthening, contralateral shortening or a combination of these procedures .

If the patient and family agree to rotationplasty and fitting as a below-knee amputee, they must be made aware of the cosmetic ramifications and the possible need for secondary procedures . **Kostuik et al.,** reported that the need for repeated rotationplasty is high (up to 50 per cent) in children under 12 years of age at the time of initial Van Nes procedure . For this reason, they recommend waiting until the child is at least 12 years old . This would also allow the patient to participate in the surgical decision making .

Patients with bilateral PFFD will usually ambulate well without prostheses . Ablation of the feet will severely limit their function and is usually not done unless other severe distal anomalies require Syme's amputation to promote prosthetic fitting . In general the patient with

bilateral involvement should be fitted with non stanard extension prostheses . As adults, these patients can then make their own decisions regarding possible ablation ***(Grogan et al., 1987)*** .