



# POSNA

## The Core Curriculum

### **Proximal focal femoral deficiency**

#### **Objectives**

1. Define proximal focal femoral deficiency (PFFD)
2. Describe a useful classification system for PFFD
3. Describe the spectrum of severity of PFFD
4. Describe current treatment approaches for PFFD

#### **Discussion**

Proximal focal femoral deficiency (PFFD) is a term used to describe a condition where the femur is short and associated with an apparent loss of continuity between the shaft and neck. Some consider the congenital short femur to be in the spectrum of PFFD; the congenital short femur does not have a loss of continuity between the shaft and neck. There are several classifications of PFFD. The Aitken focuses on the status of the femoral head and neck; A denotes the presence of a proximal femur that usually fuses to the shaft at skeletal maturity, B denotes a well defined acetabulum, but an unossified femoral head at birth, C denotes no femoral head and a poorly defined acetabulum, and D an extremely short or absent femur. Hamanishi described 10 gradations. Gillespie's is most simple and applicable. In group I, the femur is 40-60% shorter than normal, and the hip and knee can be made functional. In group II, the femur is shorter, and the hip or knee cannot be made functional. The clinical appearance of PFFD is characteristic, with a very short, fleshy thigh in a flexed, abducted and externally rotated position. The foot usually is at about the level of the normal knee. Fibular hemimelia accompanies PFFD in almost one half the cases.

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. For children who have no hip joint and a small femur, the femur has been fused to the pelvis to allow the knee joint to functionally substitute for the hip joint. For patients with a stable hip and very short femur, Symes amputation and knee fusion (with or without epiphyseodesis) allows prosthetic fitting similar to that for a knee disarticulation. The Van Nes rotationplasty essentially reverses the plane of the ankle joint at the knee level, which allows the limb to function more as would a below knee amputation. Disadvantages to the Van Nes procedure are its cosmetic appearance and the tendency for the ankle to spontaneously derotate to its original position. Recent modifications of technique have reported less derotation than previously reported. 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.

Patients with congenital short femur characteristically have an externally rotated leg secondary to femoral retroversion, hypoplasia of the lateral femoral condyle (with or without fibular hemimelia), absence of the cruciates, and an exaggerated anterolateral bow of the femur. Musculotendinous contractures also render lengthening difficult for congenital short femur. In addition to lengthening, correction of rotational and angular deformities are prerequisites to a successful result for congenital short femur. The presence of postaxial deficiencies below the knee may complicate management.

Children in developing countries 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 counseling for children with limb deficiencies is also difficult, as predictable genotype-phenotype relationships have not yet been established.

## References

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