What is Proximal Femoral Focal Deficiency?

Proximal Femoral Focal Deficiency (PFFD) and Congenital Short Femur, are congenital (present at birth) conditions. Proximal means closest to the area of attachment to the body and focal refers to the point of growth. The long bone in the thigh is called the femur; hence Proximal Femoral Focal Deficiency relates to a problem with the growth plate in the thighbone, close to the place where the hip joint is. Although often divided into the two groups above, cases of major congenital shortening of the femur range gradually from a normally formed but short femur to an almost absent femur.

In Congenital Short Femur the femur is short (on average 10% shorter than it should be) and the thigh is often bulky. The hip usually has a deformity that results in the leg facing a little outwards, rather than forward, and the knee is a little unstable.

In Proximal Focal Femoral Deficiency, both the femur and the hip are affected. There is usually major shortening - sometimes the foot is level with the knee of the other leg. The more severe the case, the shallower the angle between the top and shaft of the femur. The knee usually has some instability and the leg between the knee and ankle is often also short. The thinner of the two bones here (fibula) can be missing or underdeveloped (Fibular Hemimelia). In these cases, there may be associated foot abnormalities.

Why does it happen?

The condition is rare, occurring in approximately 1 in 40,000 births. It is usually unilateral (affecting one leg) but can be bilateral. The condition is not thought to be genetic and the only definite cause noted to date has been thalidomide. Possible causes include: lack of oxygen to the developing baby; physical injury during pregnancy; bacterial toxins; viral infection; exposure to chemicals; and hormonal problems. The condition is thought to begin in early pregnancy, around 4-6 weeks, and causes a problem with lower limb (leg) bud formation.

Diagnosis and classification

Many cases are likely to be detected at scans during pregnancy, as the thigh bone is routinely measured, although more mild cases can be missed. PFFD is classified into different groups, using Atkins’ classification, based on examination by X-ray and different treatments can be applied according to severity:

Type (A):
- The femur is short with a shallow angle between head of the thighbone and the shaft with sideways bowing in the top third.
- The acetabulum (hip socket) is unaffected.
- A gap in bone growth at the neck of the thighbone causes stunting of the bone.
- The small gap in the thighbone will eventually solidify, but the angle between the head of the thighbone and the shaft is very shallow.

Type (B):
- The hardening of the ball joint is delayed and the socket is slightly misshapen.
- A gap in bone growth at the neck of the thighbone causes stunting of the bone.
- The small gap in the thighbone will eventually solidify, but the angle between the head of the thighbone and the shaft is very shallow.

Type (C):
- The hip socket is very misshapen and the ball of the joint will not solidify.
• The thighbone is very short and its upper end tapers sharply to a point.
• The hip is very unstable.

Type (D):
• Both hip socket and ball (femoral head) are missing.
• Only the lower part of the femur has formed properly and is significantly stunted.

How can it be treated?
Predicting the leg length discrepancy at skeletal maturity is a first step to evaluating a treatment plan. This is done by X-rays of both legs to measure the lengths of all the bones in the legs. A multiplier calculation is then used to calculate the predicted leg length discrepancy. Treatment depends on the severity of the condition and stability of the hip joint:
• In mild cases the child will walk with a slight limp but in severe cases they will not be able to walk without a leg extension or an orthosis (brace).
• In children with less severe thighbone shortening, a reasonably stable hip joint and a normal foot, the hip can be stabilised further with surgery. In addition, the affected leg can be lengthened and, possibly, the unaffected leg shortened.
• In cases associated with Fibular Hemimelia, it is still possible to lengthen both bones in the affected limb, if the hip can be stabilised with surgery and the foot is functioning.
• If the only solution is a prosthetic extension to the leg (artificial limb), the knee can be fused and the foot amputated to produce a stable stump so an above-knee prosthesis can be fitted.

When will treatment start?
Once a child starts to cruise or walk, a shoe raise or lift is added to the bottom of their shoe to minimise the limb length discrepancy. Children with PFFD will usually learn to walk either at their normal age or with a slight delay, as there is no neurological barrier to learning to walk. The type and timing of surgical intervention will depend on many factors and will be discussed in detail with the child’s medical team at regular consultations. If amputation is the only option, this will usually occur after 10 months of age, with a prosthesis being fitted shortly afterwards. Knee fusion surgery happens a little later, around the age of 3-4 years. If leg lengthening and any shortening is to be considered, this usually starts a little later, around the age of 7.

How will this affect my child?
The emotional and physical effects of living with - and having treatment for - PFFD will vary with the type and duration of treatment, from child to child and family to family. As the Paralympics demonstrate, even amputation is no barrier to achieving full potential. Access to new technologies and advanced prosthesis means that your child should be able to participate fully in almost any activity.

Sources of support
Our helpline (01925 750271) is open from 9am until 5pm on weekdays, for any questions you may have about practical support. Medical enquiries will be passed to our panel of NHS consultants. Email info@steps-charity.org.uk with a specific request or fill in a contact form on our website.

The Steps closed Facebook Group https://www.facebook.com/groups/1730693800550369/ is a friendly and safe way of discussing your worries with other parents, sharing tips and finding emotional support.

Our Family Contact service identifies someone else who has been through a similar situation and who’s happy to talk about their experiences, on a one-to-one basis, to offer support and encouragement.

steps-charity.org.uk
Helpline: 01925 750271 Email: info@steps-charity.org.uk

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