Steps is the national charity working for all those whose lives are affected by childhood lower limb conditions

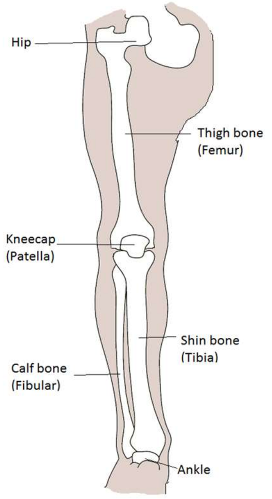
Steps
We don't take walking for granted

Fact Sheet

Proximal Femoral Focal Deficiency (PFFD)

What is PFFD/ Congenital Short Femur?

There are three main long bones in the leg: the longest is the femur (thigh bone); of the two bones in the lower leg, the thicker one is the tibia and the thinner one is the fibula.



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.

The knee is usually unstable and the leg between the knee and ankle is often also short. The thinner of the two bones below the knee (fibula) can be missing or underdeveloped (Fibular Hemimelia). In these cases, there may be also be foot problems.

Generally, in all cases, the thigh bone is short, the knee can be unstable and the leg turns outwards. Some cases have a normal hip, others do not. In some cases, the lower leg is also affected. It is problem with the way the leg develops but the exact cause is unknown and it is not usually passed on in families.

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 be genetic (passed down in families). The condition is thought to begin in early pregnancy, around 4-6 weeks, and causes a problem with lower limb (leg) formation.

Diagnosis

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 and are picked up at assessment after birth or later.

Treatment

Treatment depends on the severity of the condition and stability of the hip joint.

X-ray and other examinations will be used to try to work out what difference might develop in the length of the legs as your child grows. Treatment may include splints, insoles, or adjustments to shoes. Surgery can be used to improve the shape of the hip if needed and to consider lengthening of the leg or slowing down growth in the longer leg

- 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 so an above knee prosthesis can be fitted.

Generally, if the leg length difference is not too great, most surgeons would try and equalise the leg lengths. If the affected leg is shorter than that other types of surgery, followed use of an artificial limb are more common.

When will treatment start?

This is very much tailored to each individual child and decided upon after thorough discussion with the medical team. A consultant will usually observe a child's development to see what they can achieve on their own.

Once a child starts to cruise or walk, a shoe-raise is added to their shoe to reduce leg length discrepancy.

Children with PFFD will usually learn to walk either at their normal age or with a slight delay, there is no reason why a child with PFFD will not learn to walk. The type and timing of surgery 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 it affect my child?

The emotional and physical effects of living with - and having treatment for – PFFD will vary with the type of treatment, from child to child and family to family. Discussing your child's condition /treatment with them openly and in simple terms, promoting positive body image in both them and yourselves as parents will help them prepare for whatever the future holds.

As the many achievements of the children whose families we have supported demonstrate, even a severe case is no barrier to success and the most important thing is to always encourage full participation in whatever your child shows an interest in. Access to new technologies and advanced prosthesis means that most children 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 to 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 is a friendly and safe way of discussing your worries, 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.

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