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

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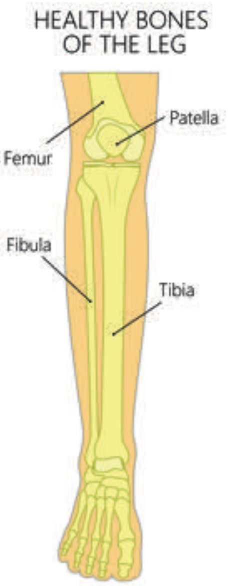
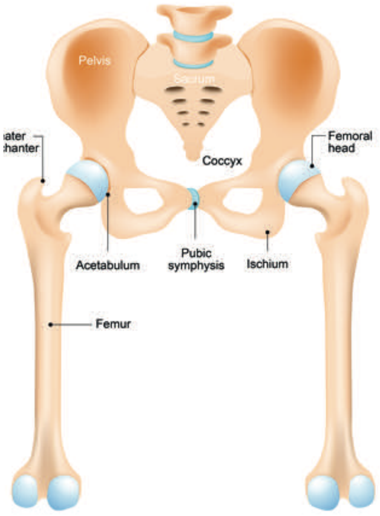
Fact Sheet

Proximal Focal Femoral Deficiency (PFFD)

What is PFFD / Congenital Short Femur?

PFFD, Proximal Femoral Focal Deficiency, Congenital Femoral Deficiency (CFD) or Congenital Short Femur, is a rare, non-hereditary, congenital (present at birth) condition which affects the hip and femur and often the knee. There are three main bones in the leg: the femur (thigh bone); tibia (shin bone) and fibula (calf bone). Of the two bones in the lower leg, the tibia is the thickest and the fibula the thinner.





The thinner of the two bones, the fibula, can be missing or underdeveloped (fibular hemimelia). PFFD, is a problem with the way the leg develops but the exact cause is still unknown, and it is not usually passed on in families.

Generally, in all cases, the thigh bone (femur), which runs from the hip joint to the knee, is short or even absent. The proximal femur is the end closest (proximal) to the hip bone and includes the rounded femoral head, which forms part of the hip joint, and the femoral neck on which the head sits (acetabulum).

With PFFD, the knee can be unstable, and the leg may be turned outwards. Some cases have a normal hip, others do not. In some cases, the lower leg is also affected.

In approximately 85% of cases of children with PFFD, the condition will only affect one leg. However, some children, up to 15% with the condition, may have bilateral PFFD which affects both legs.

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 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.

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The Leading Charity for Lower Limb Conditions
Registered charity in England and Wales (1094343), Scotland (SC049759) and also operating in Northern Ireland. A company limited by guarantee, registered in England and Wales company number 04379997.
Registered office: The White House, Wilderspool Business Park, Greenalls Avenue, Warrington, WA4 6HL.

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-rays 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 this 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. We have many inspiring family stories on our website, covering many conditions and treatment pathways at steps-charity.org.uk.

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 +44 (0) 1925 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 www.stepsworldwide.org

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 is happy to talk about their experiences, on a one to one basis, to offer support.

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