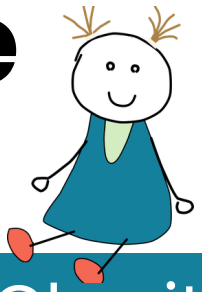
steps

We don't take walking for granted

Proximal Femoral Focal Deficiency (PFFD), a Parent's Guide



Helpful Information from Steps Charity

How This Booklet Might Help

At Steps, we know that a leg condition (often called a lower limb condition) can make us feel different and that's ok!
It can make us feel a lot of things, happy and sad, or it can confuse us and our families. This little booklet is made for children who have Proximal Femoral Focal Deficiency, their parents, or any caregivers who need more information.

This little book can't tell you everything, and it can't tell you the future, but we hope it helps. It includes medical information, helpful tips, and Grace's story of PFFD.

Proximal Femoral Focal Deficiency (PFFD) might sound like a lot of medical words, but Steps want to be your helpers and friends during this time. We're here to make things a little easier for you and your family.

We understand that sometimes you might feel a bit worried or unsure, and that's okay. We work with doctors and researchers to learn more about how to help kids with PFFD.
We want you to know that we're here to give you lots of information and bucketloads of support.

If you have questions or if you have any worries, we're here for you. We have lots of ways to support you and put you in touch with other families who have been where you are now.

Remember, you're not alone. We're going to be here with you, helping you understand and feel a bit better.

Help When You Need It

Sometimes when things feel a bit tricky, it helps to talk to someone who understands. At Steps, we have a lot of ways to help the whole family. We have a **Family Contact Service**, which means we can connect parents (or adult caregivers) with other families who have been through something similar. They can share advice, be supportive, and give helpful tips.

If you ever feel a bit unsure or have questions, adults can contact the Steps team. It's an amazing team who know a lot and can help you with all your questions. Parents and caregivers can call them at **01925 750271** or send them an email at **info@steps-charity.org.uk**. Calls are confidential so the team can help with any worries and will keep everything private.

We also have a special group on **Facebook**. There you can talk about your questions and problems with other parents. It's a safe and friendly place to share your feelings and get support and advice.

Remember there are no stupid questions, if you have a worry or a question it is important to ask.
No matter how big or small your concerns are, give us a call, send an email, or start by following us on social media.
Steps is here to help you!

Top Tip for your Grown Up:

Talk to other parents of children with PFFD, this can help you feel confident about the future and discuss things that helped them.

Visit the Steps Website at www.steps-charity.org.uk and click 'Get Help' and we can point you in the right direction.

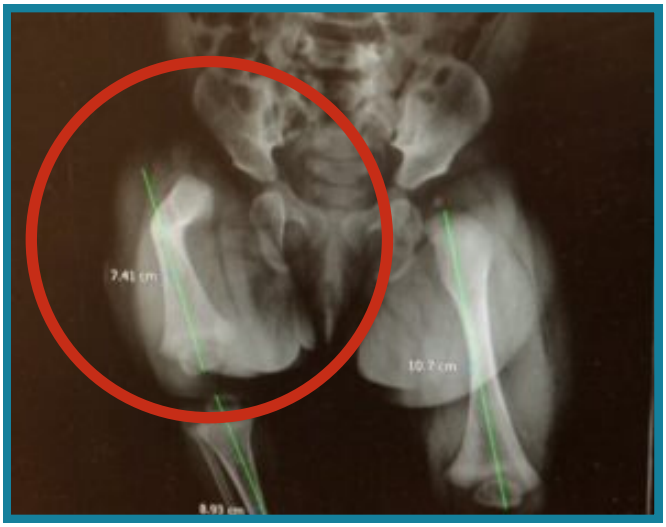
What does PFFD mean?

To understand PFFD we need to learn a few words that doctors and researchers use:

Proximal	This means "nearby" and in PFFD we're talking about the part of your leg nearest to your belly button, right at the top	..Top of the...
Femoral	If you touch your leg, you can feel the bones in there, the one at the top is called the femur, connecting your knee to your hip. Anything happening to this bone is called femoral.	...Thigh bone...
Focal	This means it happens in one part of the body. (The top of the thigh bone in your case).	...In the...
Deficiency	Something shortened or where there is not enough of something, like bone, or another type of cell.	...Shortening...

Putting this all, together PFFD means:

A shortening in the top of the thigh bone



NB: Some doctors, and some text will use the term 'Congenital Femoral Deficiency' (CFD). This is a broader term which includes PFFD and growth issues of the whole leg bone (femur) and knee.

What's happening inside my body?

For you, or your child, the first noticeable effect of PFFD will be a leg length difference, with one side being shorter than the other, this is called the **affected side**.

Every child is different and PFFD leg length difference can be small or large, you may not see any differences for a long time.

To understand why the bone is shorter we need to understand some more words:

Cartilage

A soft material that can harden to become bone, although it stays soft and squidgy in some places like the end of your nose.

Growth plate

Found at the ends of bones, growth plates are made of special cells that create cartilage. Usually, as you grow, the growth plate keeps making more cartilage, which turns into bone. This helps your limbs grow to their full length as you get older!

The growth plate at the top of your thigh bone doesn't work the way it should and so doesn't produce enough cartilage, and the cartilage it does produce might not turn into solid bone. This can mean your thigh bone might be a different shape and length on one side of your body than on the other side.

How Can Doctors Tell if I Have PFFD?

Doctors look for a few things to see if someone has PFFD. They check if one leg is shorter than the other or if the hip doesn't move as well. Here's how they can find out at different times:

Before Birth.
Sometimes, PFFD can be spotted even before a baby is born! Doctors use a special machine called an ultrasound to take pictures of the baby's bones while they're still in the belly. This helps them measure the leg bones.

Shortly After Birth.
If the difference in leg length is small, it might not show up in those scans. After the baby is born, doctors do a check called the Newborn and Infant Physical Examination (NIPE). They aren't looking for PFFD specifically, but they might notice if one leg looks different.

Later On.
Sometimes, PFFD isn't noticed until the baby grows a bit and the leg difference becomes easier to see. When that happens, doctors will take a quick and painless picture of the bones with an X-ray. This helps them see how the bones are growing. This may lead to other tests such as MRI (Magnetic resonance imaging) which your doctor will discuss with you.

In short: Having PFFD means that the part of the bone that causes bone growth doesn't work the same way in one leg as the other, so you will have one leg that grows more than the other.

Types of PFFD

PFFD can look different for different children and the doctors may tell you that your PFFD has a specific classification, this is just a fancy way of grouping similar experiences of PFFD together.

Not all consultants will tell you your type, but you are always entitled to ask as they will often use it to help communication with other clinicians about your child's needs.

There are different ways of categorising, but the "Paley" and "Aitken" categories are the most commonly used and are very similar. Below are the Aitken catagories; A, B, C, or D, and what they all mean.

Type A

Your thigh bone (femur) is a little shorter, but your hip joint is still mostly working and moving in the way that it should.

Type B

Your thigh bone is shorter, and the top part of the bone doesn't fit perfectly into the hip joint. This means that some of your hip movement is also affected.

Type C

Your thigh bone is more underdeveloped, and the hip joint doesn't really connect to it well. This can make it difficult for you to walk.

Type D

Your thigh bone didn't form properly, and there's no connection between the bone and the hip joint. Walking can be very difficult without a prosthetic leg or support.

What Can the Doctors Do?

What the doctors recommend will depend on things like what type of PFFD you have, how your hip is working, and how much it is affecting you day to day.
Doctors will talk with your parents or carer about what the options are and what might be the best treatments. Usually, they will recommend a combination of treatments and not just one.

PARENTS: Remember that you are always entitled to ask for a second (or additional) opinion from another consultant to help you make the right choice for your child and most consultants will encourage you to do this.

Observation: The Doctors may decide that the best thing to do right now is to wait and see how the limb grows, you will be asked to come and see them regularly.

A shoe raise: These may be suggested as children begin standing and walking or cruising (using furniture to hold themselves up as they walk), if they experience limitations. A raise, or extra thick sole, is used to even out the leg length.

Prosthetic leg: The doctors might send you to a limb center where someone called a prosthetist will make you something that fits over your shorter leg so that it is the same length as your longer leg. You will need new prosthetics as you grow and as your legs change.

Surgery: Surgery might be used to try and make your leg longer, make it easier to wear a prosthetic leg, or you might need surgery for your hip.

Hip Surgery: It is common in PFFD for your hip to need stabilising. The surgeons will try and make the top of your long thigh bone (the Femur bone) fit better inside the socket in your hip.

If hip surgery is right for you then after surgery you might need to wear a **hip spica cast**, just while you are healing. There is more information about these in the next pages and on the Steps website.

Leg lengthening surgery: If the difference in length between your legs is not large doctors can recommend leg lengthening surgery, they will try and lengthen the bone. Many children will need more than one leg lengthening as they keep growing. This operation involves stimulating bone growth by making small openings in the bone and slowly lengthening over time. You will go home with a fixator (something that fixes the bone in place), this can either be one you can see on the outside of the leg (external) or one inside that you can't see (internal):

External fixator -This involves a device or frame on the outside of the body, with rods connecting to the bone on the inside that have to be turned a tiny amount every day to help the bone grow.

Internal fixator - A telescopic metal rod (one that gets longer) is placed in the bone and slowly lengthens over time.

Reconstructive Amputation **:** The need for amputation and the level of amputation would be discussed with your doctor. Amputation is a surgery where they take away part of your body, in this case part of your leg. This would be to make it easier for you to walk or for a prosthetic to fit.

Rotationplasty: Although not commonly used in the UK, rotationplasty (e.g. Van Nes Rotationplasty) is sometimes suitable to those with PFFD. Its a big operation, but if the foot on your shorter side is at the height of you knee, surgeons might be able to turn the foot around so that your ankle becomes your knee joint to use in a prosthetic leg.

What is a prosthetic leg?

A prosthetic leg is a leg that is made by a prosthetist, specially for you, out of metals and plastics and other material. They can all look different for different people but they will usually look like a big boot or like your long leg.

Socket: The top of the prosthetic leg, is called a socket, this is the part that you put on, and it will be exactly the right size and shape for you to fit into. There are lots of different types of socket, depending on the shape of your leg

Children with PFFD might get an extension prosthetic, sometimes called a foot-on-foot prosthetic. Whilst children who have an amputation might have a standard prosthetic leg.

A **standard prosthetic leg** will have a socket which fits snuggly over your amputation (residual limb). You might have prosthetic knee built into the leg or you might just use your own knee.

An **extension prosthesis** will have a socket which ends in a foot shape and might have open toes. This part is strong and supports your foot so you can stand up straight and walk. Inside, it often has soft parts to make it comfy. Underneath your real foot will be a prosthetic foot, which touches the ground and helps you move.

To make the prosthetic the perfect fit, the prosthetist will take some very careful measurements of your shorter leg. This is a painless process using either a laser measure or plaster of Paris wrapped around your leg. They might let you pick a colour for your leg! Remember that this leg won't grow with you, so you will need a new leg and another visit to the prosthetist regularly to keep up with your growing body.

A new leg can feel a bit funny to start with, but a physiotherapist will help teach you how to move and walk with your new leg.

You may also need special legs to help you with activities like running around, or going to the swimming pool. This might be what we call a blade, or might be another sports limb. These are available for all types of prosthetic sockets, talk to your clinical team.

What is a Hip Spica Cast?

A hip spica cast is like a covering for your hip. It's made from strong materials, similar to what you see when someone has broken their arm, a mixture of plaster of Paris and fibreglass. You'll see lots of white ones, but sometimes they can be coloured and patterned!

Think of it as wearing stiff trousers, but each pair is different, depending on what you need. It can extend from above your belly button all the way down to the ankle, or it can be shorter; there might be a bar across the middle or nothing at all. The cast will usually hold your poorly hip in a fixed position, wider than before, and help your recovery.

It can feel a bit funny at first, but you should get used to it. To help you go to the toilet there's an opening like a little window in the front and back, these should also help nappy changes. If you don't wear nappies you may still need to use a bedpan (something that looks like a toilet seat with a container that can slip under you in bed) or a special bottle to urinate (wee) into.

This can be difficult to get used to, but the equipment is designed to make it as easy as possible. Now, because of this stiff covering, you might need some different clothes that fit over it and, when you ride in a car or a pushchair, you might need a bit more room. Don't worry, the Steps community can help you find carefully designed clothes and equipment that will accommodate the cast.

The cast is there to help your leg and hip get better after surgery. If you ever need something specific or have questions, just let the people helping you know – they're here to make things as comfortable as possible for you and remember you can reach out to Steps for advice at any time.

You won't wear the cast forever; you might even need it replaced a few times as it starts to get dirty and wear around the edges. Most clinics will only keep you in a cast for a maximum of 12 weeks.

What if surgery is recommended?

Remember that treatment doesn't always mean surgery, especially as different countries have different processes. It can be a combination of treatments, and it might mean you are asked not to walk (bear weight) for a while.

If the doctors do recommend surgery, it may be a decision for Mum, Dad or your caregiver. Surgery can be a big decision and it is worth asking your doctor why they think this is the best course of action. Make sure they are clear about possible long-term complications of both performing and not performing the surgery. Some doctors will be open to trying less invasive methods (like surgery) first if this is what the family prefers. If you find it difficult to talk to your doctor, try and find a team member who can help you communicate, or find a consultant you can build a relationship with.

Talk to the Steps community, your own family, other consultants, and do your own investigating to understand the research which is available.
The final decision is up to you as a family and the more information you have the more informed a decision you can make.

TOP TIP

Ask your consultant questions about:

- Why they recommend a treatment
- What treatment options are
- What the likely outcomes are
- What happens if surgery is postponed until they are older
- If they have previous experience of these operations

Can I Be Active?

Wearing a prosthetic limb or having a leg length difference can mean that it takes more energy for you to run around than for your friends. For some kids the doctor might tell you to avoid jumping or hopping or things that might mean your hip is under pressure, but this will usually only be while you recover from surgery.

YES! YOU CAN BE ACTIVE!
Maybe you are a climber? Or love a game of netball? How about some gymnastics? Well, you should be able to take part in whatever activity you like and be with your friends.

If you do have any concerns or find anything difficult talk to your clinical team (doctors, physios etc). They are like your sidekicks, and they'll help you figure out the best way to keep having awesome adventures while taking care of your body.

There are also, all sorts of adapted and seated games, music and arts that you can play. Now is the time to try new activities and maybe find a new hobby for life.

Top Tip

Activity doesn't have to mean running, think of other fun things you can try.

Horse riding?
Paddle boarding?
Walking football?

If you aren't sure what activities you might like then try some taster sessions.

You always need to listen to the doctor but remember that sometimes how active you can be, and how active your friend can be, might be different.
We know this can be confusing, and you might feel lots of emotions, both bad and good, about this. It is important for the whole family to think about your mental health.

What About When I'm Older?

PFFD doesn't stop you from being amazing, children with PFFD have gone on to dance ballet, play football, and achieve wonderful careers.

PFFD is never cured and so your leg and hip will always be a bit different. As you grow, doctors will keep checking how they are growing and how well the hip and leg fit together. How things feel when you are older depends on what type of PFFD you have and how much your leg was affected in your childhood. For example If you need a prosthetic leg when you are a child, you will need an adult size one when you are grown up.

REMEMBER

PFFD is a rare condition so a lot of the research is "Case studies".

These look at one person's experience of PFFD at a time. This helps us understand the condition.

BUT,

It is important to remember that everyone is different and so is your experience of PFFD.

So find out what you can do and what works for you!

When you're older, you might still notice differences in how you walk or move, but you will have learnt to do so many things that you love!

Because you move a bit differently you might find you get aches and pains in your back and hips and what we call "wear and tear". You might need to rest more than friends and it is a good idea to visit a physio regularly and keep up some strength and flexibility training to make sure your body is as healthy as possible.

Grace's Story

When Grace was still in her Mum's tummy the Doctors noticed during a scan that Grace seemed to have one leg smaller than the other. The Doctors were a bit baffled but eventually decided she had a condition called PFFD.
Grace was born a little bit earlier than expected but came out with a big cry and her Mum and Dad knew then that she was going to be a strong girl!

When Grace was born her right leg was about 1cm shorter than the left but as she got bigger, so did this difference.

When she started walking at 13 months it was about 3cms shorter and the doctors made her a special raised shoe which she quickly got used to.

When she was 3 years old the doctors decided it was time for her to have her 'SUPERHIP' surgery. After surgery she had to wear a Spica Cast for 6 weeks. Her Mum and Dad found it very tricky to keep her entertained in this time. But, Grace liked being the boss and enjoyed everyone waiting on her.

Her nursery were very helpful and even employed a special lady to help her so she could still go in and see her friends when she felt, even when she couldn't walk.

Once the Spica Cast came off Grace enjoyed living life to the full and joining all sorts of clubs including Rainbows, Ballet and, her true passion, Gymnastics. Grace enjoyed learning cartwheels, beam skills and round-offs. She was extremely proud when she won bronze medal in her gymnastics competition.

When she started school, Grace wore a raised shoe. She always fully took part in all activities, beating 60 other kids in her class in a climbing competition on a school trip, and really enjoyed taking part in all the activities at the Steps camp. The kids in her class all knew about her tiny leg (as Grace liked to call it) but Grace did sometimes find it super boring when other kids asked her about it at playtime or when she was at the park.

Grace's doctors started to talk about the lengthening which her parents had decided on when suddenly Covid struck the world. Appointments and talk of lengthening halted and Graces leg length difference continued to grow, when hospitals started offering appointments again following Covid it was decided she would try a 'foot on foot' prosthetic, her difference was about 8 or 9cm at this point.

The waiting lists for lengthening had grown and Grace was finding her difference harder to manage, so the prosthetic leg really helped her.

She was able to wear it for gymnastics and it helped her to run even faster than before.
She wore this for a while but found lots of people were asking questions about it (some people even thought she had 2 feet on one leg!).

So, she asked if she could try a prosthetic extension blade which at the time was quite unheard of.

The specialists worked together to make her one and Grace immediately loved it - it made her very bouncy and she was even able to choose the pattern on it!

The next part of the story is a bit sad, because the Doctors found that a complication had occurred with her hip and instead of undergoing lengthening they had to perform another SUPERHIP procedure and Grace even got given a wheelchair.

But this didn't stop Grace! She quickly learnt some cool skills, she performed wheelies and her clever Dad put pink lights on the wheels.

Grace was keen to get back to her activities, and as soon as she was given the all clear she was straight out of her wheelchair and building up her skills again. After 10 months in her wheelchair she was back to walking and performing gymnastics in just 3 weeks! Grace is now 9 years old and will probably have her leg lengthened in the next few years.

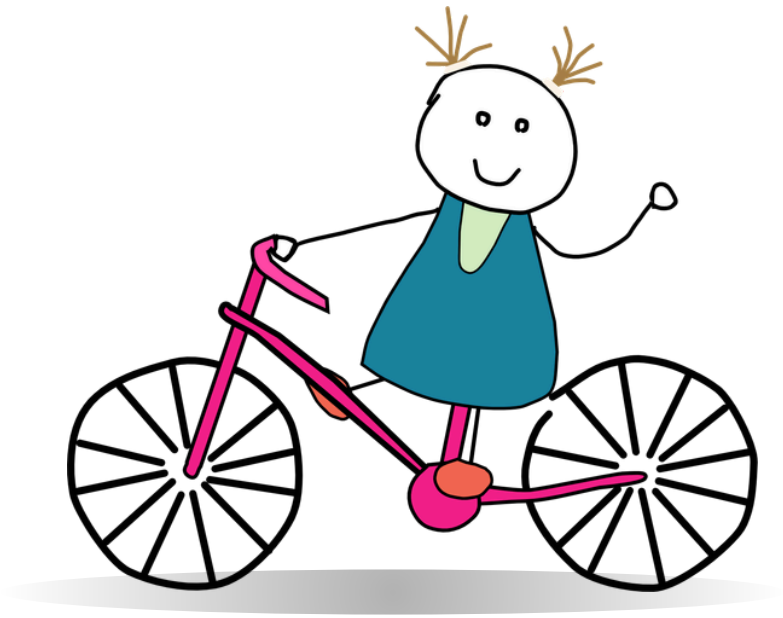
To read more stories like this, visit www.steps-charity.org.uk

What Do I Do Now?

Having PFFD can affect each person differently, depending on your age, the extent of your leg and hip development, and the resources available to you. This means that the treatment approach your doctors recommend might be different from someone else's. It's important for you, your family, and your healthcare team to work together to find the best treatment plan for you. In the UK, there aren't yet specific guidelines for treating PFFD (though researchers are working on it!). There are many options to consider, and your preferences and thoughts about your treatment are important in making the best choices for your care.

Steps can be helpful in this journey. Even though we can't predict the future, talking to other families who went through similar treatments can make things clearer and help you understand how it might affect different parts of your life. The more you know, the better prepared you can be!

After a diagnosis, or even the first indications of a problem, it can feel scary and it's normal to feel sad or worried. The whole family needs time to talk about how they are feeling, with each other and, with the medical professional. We know it's a lot of new information all at once, but Steps have decades of experience helping families like yours get the information they need to make the journey a lot less scary.



Who are Steps and How Can They Help?

Steps is the national charity working for all those whose lives are affected by childhood lower limb conditions. Everything we do is about valuing and supporting individuals, families and carers affected by conditions which have an impact on the legs, hips or feet.

Contact Steps (confidential)

Call 01925 750271 with any questions you have or email info@steps-charity.org.uk

Facebook discussion groups (private)

There are two pages for members to join:
Steps Charity - Support Group for Parents
Steps Charity - Support Group for Adults

Family contact support service (confidential)

Connecting parents (or adult caregivers) with other families who have been through something similar. Enquire via our contact number 01925 750271 or email info@steps-charity.org.uk. All our Family Contacts are interviewed and given training before they engage with a family, providing a forum for sharing ideas and consulting other parents on day-to-day questions.

Social media

Steps is active on X (@Steps_Charity), Instagram (@StepsCharity) and on YouTube (StepsCharity)

Please note, external websites are not associated with Steps and Steps are not responsible for their content.

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Steps Charity Groups:
Steps Charity - Support Group for Parents
Steps Charity - Support Group for Adults

Remember, you are not alone on this journey. Steps is here to offer a helping hand, a listening ear, and a community that understands. We believe in the strength of shared experiences, and our doors are always open for you. Whether you need information, guidance, or simply someone to talk to, you can reach out to us. Together, we can navigate the path ahead. You are part of a supportive community, and we're here to help you every step of the way.

"It was your videos with Dr. Perry that really helped me help my son when he was first diagnosed. It was because of steps and those videos that gave me clues on what I needed to look for and how best to support my son since there is no cure. I appreciated all the transparency from the videos and I still recommended them to this day."

"Steps have been an absolute lifesaver for me they have provided me with information on Mica's condition and treatment and I have been able to discuss Mica's condition with other families and read how people have coped with the long treatments"

Steps is here to help you like they have helped so many families since their creation in 1975.

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Steps Charity Groups:
Steps Charity - Support Group for Parents
Steps Charity - Support Group for Adults

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Contributors

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