Practical advice and medical information about caring for a child affected by Lower Limb Deficiency
A glimpse inside... inspiring personal stories

**Harry** was born with PFFD and underwent an amputation at 11 months old. Read his parents inspiring story on pages 24 - 25.

At 11 months old **Nick Gibbon** underwent a Symes amputation. He went on to represent Wales at the 2000 Commonwealth Games. Read Nick’s inspiring story on pages 26 - 27.

At birth, **Xander** was diagnosed with fibular hemimelia - now nearly 3 years old he is still too young for leg lengthening treatment. Read his story so far on pages 28 - 30.

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Front cover picture: **Yasmin Depass** born with PFFD
Introduction

For many parents the diagnosis of a child with a lower limb deficiency can be a shattering experience which may leave you feeling isolated and alone. Lower limb deficiency is a rare condition and an overwhelming need for good information and emotional support exists. You will probably have many questions and be asked to make decisions which you may feel ill equipped to reach.

You are not alone

STEPS is here for adults and children affected by a lower limb condition including clubfoot and hip dysplasia. We bring people together, provide dedicated information and support and campaign for improved standards of care.

Established in 1980, our valuable experience and knowledge can help you make confident and informed decisions at what can be a difficult and confusing time.

Support when you need it

Get instant access to valuable information and parents stories via our comprehensive website. You can also visit our online support forum and ‘meet’ others facing a similar situation to yourself and share problems and solutions. Being able to contact someone who knows exactly what you are going through can often be the biggest help when dealing with an uncertain situation. Our Family Contacts service is an informal support service putting families in touch with others who have shared a similar experience.

When you want information, advice and support our helpline team are here to offer an expert ear and understanding in total confidence. No matter how big or small your concern please telephone: 01925 750271 or email info@steps-charity.

It is impossible for us to know at what stage you are reading this booklet and what lower limb deficiency affects your child. No book can give all the answers required but hopefully this one will help you to gain an understanding of your child's condition and show you how to cope effectively with lower limb deficiency.

"We have moved on from deep sadness at the diagnosis to a feeling of pure joy at his existence. To us, he is of course, perfect."

Amy Colquhoun, proud mum of Euan affected by Fibular Hemimelia who underwent a below the knee amputation aged 11 months.
What is Lower Limb Deficiency?

Lower Limb Deficiency is when the leg is shortened or incorrectly formed at birth.

The most common types of Lower Limb Deficiency:

- **Proximal femoral focal deficiency (PFFD) and Congenital Short Femur**
- **Congenital Short Tibia with absent or hypoplastic fibula (Fibular Hemimelia/Dysplasia)**
- **Congenital dysplasia or absence of the tibia with intact fibula (Tibial Hemimelia/Dysplasia)**
- **Coxa Vara**

Proximal femoral focal deficiency (PFFD) and Congenital Short Femur

Although often divided into these two groups, cases of major congenital shortening of the femur range gradually from a normally formed but short femur to an almost absent femur.

**Congenital Short Femur**

The femur (thighbone) 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.

**Proximal Focal Femoral Deficiency**

- Both the femur (thighbone) and the hip are affected. There is usually major shortening - sometimes the foot is level with the knee of the other leg
- The knee usually has some instability
- The leg between the knee and ankle is often also short, the thinner of the two bones here (fibula) can be missing or underdeveloped
- The foot is often fairly normal
- The more severe the condition the less the angle between the top and shaft of the femur

**Diagnosis**

The femur is measured as part of a routine ultrasound scan during pregnancy. Diagnosis at this stage will depend on the amount of shortening. In milder cases diagnosis will only be made at birth or later as the legs start to grow and the difference becomes more noticeable.

**How can it be treated?**

Congenital short femur can often be treated by leg lengthening, but the knee and hip can become more unstable during this treatment so this needs to be thought about before a decision is made.

Proximal Femoral Focal Deficiency (PFFD) may sometimes be treated using limb lengthening techniques but often results in a deficiency that is too great, or a knee and hip that are too unstable, for limb lengthening to be successful. The hip will often need to be stabilised by surgery.

Where 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 that an above-knee prosthesis can be fitted
- The Van Nes Rotationplasty can be used – again, the knee is fused but the leg is turned to face backwards and the ankle joint is used as a knee. The foot fits into the prosthesis
THE PARENTS GUIDE TO LOWER LIMB DEFICIENCY

Congenital Short Tibia with absent or hypoplastic fibula (Fibular Hemimelia/Dysplasia)

There are two long bones in the lower leg, the thicker one is called the tibia and the thinner one is the fibula. In this condition the tibia is shorter than normal and the fibula is missing or underdeveloped.

- The tibia may be bent
- The foot may also be smaller than normal, bent outwards at the ankle and may have fewer than 5 toes
- The knee is often also misshapen and may move abnormally. The condition may affect soft tissue as well as bones
- It is the most common form of lower limb deficiency present at birth
- It occurs in approximately 1 in 25,000 births

Diagnosis
Severe cases are likely to be seen at scans during pregnancy. In milder cases it may at first be thought to be a foot deformity but when the shortening of the leg becomes more noticeable and the leg is X-rayed the full picture can be seen.

How can it be treated?
Treatment will depend on the severity of the condition and how normal the foot is.

- If the foot is reasonably normal it may be possible to lengthen the leg using distraction apparatus such as frames
- If the foot is twisted outwards to the side it may be possible to correct it sufficiently using surgery
- If the foot is not sufficiently normally formed and the tibia is too short it may be best to amputate it through the ankle using procedures such as the Syme's or Boyd amputation and fit a prosthesis (an artificial limb)
- In cases where the fibula is present and the shortening is not severe the treatment is usually leg equalisation either by slowing growth of the other leg (epiphysseodesis) or leg lengthening

THE PARENTS GUIDE TO LOWER LIMB DEFICIENCY

Congenital Dysplasia or absence of the tibia with intact fibula (Tibial Hemimelia/ Dysplasia)

There are two long bones in the lower leg, the thicker one is called the tibia and the thinner one is the fibula. In this condition the fibula is present but the tibia is either missing or mis-shapen.

- There is a foot deformity and the foot may have additional toes
- The knee may be twisted inwards
- The X-ray at 1 year old gives much more information than at birth
- It occurs in approximately 1 in 1,000,000 live births

Diagnosis
At or before birth during a routine scan.

How can it be treated?
Treatment depends on the exact form of the condition.

- For the mildest form distraction apparatus such as frames may be used to lengthen the leg and improve the position of the foot
- In more severe cases amputation is often the best option with the provision of a prosthetic limb which may be above or below the knee depending on the condition of the knee joint and the presence or absence of certain other parts of the upper leg
- Some attempts at reconstruction using the fibula have been made but rarely provide a well functioning limb due to instability at the knee and problems with the quadriceps (thigh) musculature
Coxa Vara

This is a problem with the shape of the hip. The femur is the long leg bone above the knee. It has a ball-shaped upper end which fits into the socket of the pelvis. The angle between this ball and the shaft of the femur is less than normal which results in the leg being shortened.

**Diagnosis**

Usually after walking age the parents notice that the child has a limp and one leg seems to be shorter than the other. It is also harder to move the leg outwards from the hip and the whole leg may be rotated slightly. Sometimes it is initially thought to be Developmental Dysplasia of the Hip but in Coxa Vara the changes shown by an x-ray are only to the femoral neck.

**How can it be treated?**

If the angle, which should be about $140^\circ$, is actually greater than $110^\circ$ it may not need surgery.

If the angle is less than $110^\circ$ it is likely that the deformity will worsen. A shoe raise may be used initially but the Coxa Vara will be monitored and if it decreases to less than $100^\circ$, surgery to the bone may be used to correct the angle. This type of surgery may need to be repeated, especially if it is first carried out when the child is very young but the leg length difference is rarely enough to require leg lengthening.
What causes Lower Limb Deficiency?

It is only natural to ask why someone has a condition like Lower Limb Deficiency. In the majority of cases, medical staff are genuinely unable to provide a suitable explanation.

Often the cause is unknown but it is thought that something causes damage at the crucial time that the leg is developing during pregnancy in the first trimester (between days 28 and 42). Some discrepancies are due to genetic conditions. Other possible risk factors are smoking during pregnancy, viruses, accidental damage, drugs or some kind of disruption to the local blood supply.

When is Lower Limb Deficiency diagnosed?

During Pregnancy
With the advancement of ultrasound scanning, Lower Limb Deficiency is usually diagnosed during pregnancy. At your scan the length of the femur is measured and the tibia, fibula and foot are checked. If the length of the femur is not within the normal range the other limbs will be measured.

You may be offered further tests and if so will have to decide whether to accept them. Early detection of this nature can have its advantages and disadvantages. Knowing about any problems your baby may have allows you to prepare for the birth and find out about the condition and treatment options available. On the other hand, uncertainty can still remain after testing as false-positives and false-negatives are possible. There is also a risk of about 1% of suffering a miscarriage when invasive testing is carried out.

Birth and beyond
In some cases, lower limb deficiency will only be diagnosed at birth or when your child starts to walk. The shock and anxiety felt at this time is perfectly natural and you will probably really want information and possible outcomes. In such circumstances, it may be constructive for you to share your thoughts and feelings with others who have experienced a similar occurrence in their lives. Early referral to a specialist centre is also desirable.

What treatments options are available for Lower Limb Deficiency?

Often it is necessary to wait for any surgery until the child reaches a certain age. Until that time both legs will be monitored, both in terms of the discrepancy and any signs of developing deformities. There is no need to rush into treatment. It is important to have time to think about the options.

The options for treatment will vary depending on which bones are present, the size and shape of them and any abnormalities in other structures such as muscles and ligaments. The doctor has to take into account the rate of growth of each leg and the amount of time growth is expected to continue for. Other abnormalities may also be present which may increase or reduce the apparent difference in leg lengths.

Depending on the type of Lower Limb Deficiency affecting your child there are two basic options available:
- Long term management of legs of different lengths
- Leg equalisation

Della Carey with Ilizarov Frame treatment for PFFD
Long term management of legs of different lengths

Leg length differences of less than 2cm are common and don’t usually cause any long-term problems. Problems associated with small differences can often be solved with a shoe raise which can frequently be hidden inside the shoe.

Differences greater than 2cm cause the child to stand unevenly and will affect the way that they walk. A larger shoe raise, or a specially made leg extension called an orthosis can be fitted but if the difference is expected to cause other orthopaedic problems, such as backache or osteoarthritis of the knee and hip of the longer leg, then correction of the discrepancy may be a better option.

In some cases an amputation of the foot and part of the leg may be required because the structure of the shorter leg means that it would be difficult for the child to walk without this treatment. It allows a well-fitting prosthesis to be fitted so that the child can walk despite the deficiency that they were born with. To find out more about amputation please call 01925 750271 and we can also put you in touch with families who have shared a similar experience to you.

Leg equalisation

This may be achieved by either shortening the long leg or lengthening the short leg

Shortening may be achieved by:

- **Epiphysiodesis** - surgery to the growth plate of the longer leg to stop further growth. This is appropriate when there is sufficient growth left to allow correction, the child is going to be taller than average and the difference is 6cm or less. Or if it is unsafe to lengthen the shorter leg. One form of epiphysiodesis is Stapling – inserting staples across the part of the bone that grows (growth plate), which is at the end of each of the long bones in the leg.

- **Femoral and tibial shortening** - usually femoral, and carried out at the top of the bone so that scarring and bulkiness can be hidden by clothing. This is appropriate when the patient has stopped growing, the child is taller than average, the discrepancy is mainly in the femur and the difference is 6cm or less.

Lengthening - the short leg may lengthened by procedures such as:

- **the Salter osteotomy** which inserts a wedge of bone into the pelvis
- **the use of a distraction apparatus**, which works by pulling apart a deliberate break in the bone and allowing the natural healing process to fill the gap. Techniques include: the Ilizarov frame, the Taylor Spatial frame (all round/circular fixators), the Wagner or Orthofix devices (single-sided bars) and intramedullary lengthening (albizzia nail)

Lengthening is a long term process and if your child requires this treatment please call our helpline on 01925 750271 or email info@steps-charity.org.uk

Choosing where to have treatment

When considering treatment options for your child it is important to remember there is not a ‘right’ or ‘wrong’ decision. It is based on the beliefs and aspiration of each unique family and your child’s Lower Limb Deficiency. Never feel pressured into making any quick decisions and always ensure that you have clear unbiased information about your child’s treatment plan. There are several hospitals which have paediatric orthopaedic consultants who specialise in lower limb deficiency but they may not be local to where you live. However, under the NHS free choice service you can now ask to be referred to one of these hospitals if you feel that would best suit your needs.

If you would like to find your nearest specialist orthopaedic consultant or limb fitting centre (if applicable) please call the STEPS helpline on 01925 750271 or email info@steps-charity.org.uk to ask for the details.
Communicating with doctors

It is important to understand exactly what your child's treatment will be and how it will help your child. Having a list of questions or topics that you want to discuss at your appointment will make it more likely that everything is covered.

You should not be afraid to ask questions. Parents often get a lot of information in a short amount of time during a typical visit. If you are not given a handout with the information written down, ask for one, or take notes and ask questions about things you don’t understand. If you don’t ask any questions, your doctor may assume that you understand everything.

Another way to ensure you really grasp what’s going on during the appointment is to bring a partner or friend along who can serve as another set of ears. If there are two of you there, you’re more likely to get a balanced perspective. You can discuss what you heard afterwards. Also, everyone forgets a certain amount of what a doctor says - having another person there can help you remember key facts.

Getting a second opinion
If you would like to consult with another expert about your child's treatment plan you may ask for a second (or further) opinion. Getting a second opinion should not offend the original doctor. Before asking for a second opinion, it is worth asking your doctor to go over and explain anything you do not understand. Although you do not have a legal right to a second opinion, you do have the right to ask for one.

If you have any questions or need assistance regarding getting a second opinion please call our helpline 01925 750271 or email info@steps-charity.org.uk.
Your feelings and fears about the future

You may just feel generally worried or may have more specific concerns about your child's Lower Limb Deficiency. Lacking experience in dealing with your child’s condition you naturally do not know what to expect.

Until you see your child develop some natural abilities it is possible you may under-estimate their potential. To reassure you, many of the parents who have contacted STEPS have said that the development of a child with a Lower Limb Deficiency is similar to the development of any other child.

Remember, none of us can predict or control the future; you can only affect the present. Like all parents, you will find yourself pleasantly surprised as you watch your child grow.

Reaction of other people

Parents are often concerned about how other people will react to their child and often worry about handling it. Actually, there is no magic formula or one ‘right way’. You will find through time, contact with other STEPS families and by personal experience the way that is comfortable and right for you and your child.

It is important that your child develops a healthy attitude and self acceptance about his Lower Limb Deficiency. Positive parents are more likely to have positive children with high self esteem. If your child realises you accept the condition without embarrassment, it will help him accept himself in the same way and deal with any negative attitudes. You can do this by allowing him to take advantage of many opportunities for self development and responding as best you can to your child's needs and feeling.

You will discover that your child's needs are the same as they are for all children regardless of his lower limb condition. Generally once people see that you don't treat your child any differently they too will learn to accept your child as just another little boy or girl.
Your child’s perception

At some time in most children’s lives, whether they have a Lower Limb Deficiency or not they will go through a stage of self consciousness. Physical issues like appearance and clothes can become very important.

It is at this time that some children begin to try to hide their deficiency, even though they have accepted it in the past. They may for the first time show signs of resentment and feel a sense of injustice - why me? They are striving to make sense of themselves, and if there are problems, then calm and sympathetic discussions with parents or close friends and family can help them through this time.

Children’s personalities vary, and some children ask questions, while others don’t. Show your child that you are open to questions and if they arise try and answer them in as honest and sensitive a way as possible. Your child will listen to how you describe their condition to other people and probably copy that explanation, so being matter of fact and using simple terms will pay dividends.

Education

The majority of parents with a child affected by a Lower Limb Deficiency find that they may have a few problems with their child attending mainstream school. It is advisable to explain your child’s condition to the playgroup leader or head teacher before the child starts school so that any anxieties and potential problems can be identified beforehand.

Most schools are sympathetic and accommodating but you may encounter problems and you may feel that your child will need extra help or adjustments to their environment at school. Your child’s rights are outlined below:

Disability

The Disability Discrimination Act 1995 (DDA) and the Disability Equality Duty which was introduced into it in 2005 set out the provision schools should make for disabled pupils.

The DDA defines a disabled person as someone who has:

‘a physical or mental impairment which has a substantial and long-term adverse effect on his or her ability to carry out normal day-to-day activities.’

Each school should publish their disability equality and accessibility schemes as separate documents or as part of other school documents and must be able to provide copies to anyone asking for them. Schemes had to be published by 3 December 2007 and schools are required to report on them annually, so when choosing a school you should be able to compare provision by asking for copies of these schemes and the school’s reports on their own progress.

There is a significant overlap between children and young people who count as disabled under the DDA and those who have special educational needs as defined by the Education Act 1996. The definition of SEN includes many, but not necessarily all, disabled children: a disabled child has special educational needs if they have a disability and need special educational provision to be made for them in order to be able to access the education which is available locally.

Special Educational Needs

A special educational need is defined as,

‘some kind of sensory or physical needs which may affect them in school’

If your child has more difficulties than most children their age with their schoolwork, communication or behaviour, you are entitled to help from special educational needs specialists, teachers, and voluntary organisations.

Initially you should discuss your child’s difficulties with the school’s special educational needs co-ordinator (SENCO), teacher or head teacher but if, at any time, you are not satisfied with the provision for your child there are other sources of help and advice such as your local parent partnership service which can provide, ‘accurate and neutral information on the full range of options available to parents.’ However you may find that you have to be very proactive in sourcing this help for your child.
Claiming Disability Living Allowance (DLA)

If your child has care or mobility needs due to an illness or disability you may be able to claim Disability Living Allowance for them. It doesn't matter if you don't consider your child to be 'disabled'. For the purposes of DLA, 'disabled' simply means that your child has a long term condition which affects their everyday activities. It also doesn't matter if you've been told by anyone (including the Department for Work and Pensions, your GP, a nurse, social worker etc.), other than a professional welfare rights worker, not to make a claim because your child is not disabled enough. The question of entitlement is a legal one, not a medical one. If in doubt, make a claim.

What is DLA?
Disability Living Allowance is made up of two components - care and mobility.

If your child has personal care needs at certain parts of the day they may be entitled to the low rate care component. The middle rate of care is paid to children who need care during either the day or night and the high rate of care is paid if they need care during both day and night. To claim you must be able to demonstrate that these needs are substantially more than those of healthy children of the same age.

Disability Living Allowance mobility component is paid if your child can walk but needs assistance and the high rate of mobility is paid if they have great difficulty in walking, cannot walk at all or are deaf and blind and need assistance to get around.

Am I eligible to claim DLA for my child?
If your child has an illness or disability that affects their daily life then it is worth finding out whether you can claim Disability Living Allowance for them. Each claim is assessed individually to see whether the eligibility rules are met and which rate is payable. Disability Living Allowance for children is not means-tested so it does not matter what other income or savings you have.

Your child must be aged 3 months or over to claim care component. To claim mobility component they must be at least 3 years old.
Better than expected

When Lisa Cockcroft's son Harrison was born with PFFD they discovered the best treatment option would be an amputation through the ankle. Read Lisa's emotional story of how she began to realize that Harry's condition wasn't as bad as she had first feared.

We found out that Harry had a short femur at our 20 week scan. It came as a complete shock after already having a perfectly healthy baby the year before. We were asked if we would like an amniocentesis to see if a chromosome defect had caused the short femur. We declined as we didn't want to risk miscarrying the baby that we already loved with all our hearts. We knew that the results wouldn't make his leg grow so it wasn't a risk we were prepared to take. We went for a more detailed scan at Leeds UGI, where all Harry's bones were measured and compared. Everything else looked as it should and the short femur seemed to be the only problem. Nothing of this nature ran in either of our families so they didn't feel it was a genetic condition. After this appointment we were put in touch with Mr Fernandes who is a paediatric orthopaedic surgeon at Sheffield Children's Hospital and would discuss all our options with us.

While we waited for our appointment, I started to carry out research on the internet. We knew the femur was hypoplastic and I found lots of research leading to a condition called Proximal femoral focal deficiency (PFFD) which described several treatment methods, depending on the length difference. We already knew from our detailed scan that Harry's right femur was shorter by around 55%. The research I found on this length discrepancy was very upsetting.

Amputation sounded like such a drastic measure but unfortunately a likely outcome for our unborn baby. I also came across the STEPS web page, for months I just read other people's stories and posts on the forum, and I began to realise that Harry's condition really isn't that bad. I was amazed at how well other babies and children coped and at how strong their parents were but when they did need support and guidance members of STEPS were always there.

Harry was born right on due date, 12th June 2008 by normal delivery. He was beautiful. When Harry was 3 months old we had an appointment to see Mr Fernandes, who examined Harry and carried out some x-rays on both legs and hips., and concluded that Harry's best option would be amputation at the age of 11 months. We had many appointments over the next few months to prepare ourselves and ask all the questions we needed to. And in this time Harry grew into a happy cheeky little boy, and even with a short leg it didn't stop him pulling himself up to stand or crawling at lightening speed.

In March 2009 we were given a date for Harry's operation for 14th May. It was such a relief to have a date to prepare ourselves for. Unfortunately Harry caught a cold the day before and his operation had to be postponed, I can't even express how down I felt when we just wanted this hard part over. Luckily we got a new date but I was on edge the whole time until he went for his surgery.

The amputation was a great success. Dye was also injected into Harry' hip so that Mr Fernandes could get a good look at the ball, socket and mobility. Thankfully the hip joint has formed perfectly but the mobility isn't great so at around the age of 2 Harry will need some hip reconstruction carried out, but that's something to worry about next year. Harry recovered amazingly well from the amputation and was crawling around again in a matter of days. Even the day after the operation he was pulling himself up at the side of the hospital cot. We saw Harry's stump for the first time the day after the operation and we were amazed at how well it looked and it was not at all as scary as we through it would be seeing it for the first time. On the fourth day we were allowed home.

Our first night went really well with Harry only waking through the night when he was due his medication. The days were more difficult because Harry never stopped still and was unwilling to go in a baby walker his bandages would work their way down his leg and come completely off. On our first day at home we had to go to our local hospital twice to get his leg redressed. We were fast learners and on the second day we were able to dress his leg ourselves. We are now very used to seeing Harry's leg without a foot on the end of it.

In about 6 to 8 weeks Harry will go to have measurements taken to get his first prosthesis. It will be a very basic leg that will encase his knee so that he will walk with more of a swing of the hip. As he gets older Mr Fernandes will assess how Harry copes with his knee and if he feels Harry will benefit from having his knee fused together this will be carried out at a later stage. We knew from the very first day Harry started treatment that we made the right choice as Harry has shown great determination to carry on like the amputation never happened. We are so excited to be getting his first prosthesis soon, as we know it won't be long before he is walking and running around after his big brother and friends. It is thanks to our friends and family that we have stayed strong and have gotten through the first stage with flying colours.
Where there’s a will there’s a way

Having a lower limb deficiency doesn’t mean you can’t lead a very full and active life as Nick Gibbon has already proved at just 23 years old. Read an adults perspective of how an amputation on his lower right leg hasn’t affected his ability to excel at various sports and achieve academic success.

When I was born I had a very abnormal right lower leg and foot, I was missing one of the vital lower leg bones (fibula missing and a short, bowed tibia). When I was 11 months old I had a Symes amputation on my lower right leg at the Nuffield Orthopaedic hospital in Oxford. My consultant is Mr Mike Benson and I also attend the limb centre there for my prosthesis. My prosthetic team has always been very supportive and worked hard to give me the best possible limbs. Marie Kane is my prosthetist and she has been for 15 years. I used to visit the limb centre every 3 months but in the last 5 years I have been 5 or 6 times, mainly to have new feet put on, seeing as I seem to break them every 12 months with my active lifestyle.

I have had a few operations in my time; arthroscopies due to an injury sustained playing rugby and epiphyseal stapling (a standard procedure for my abnormality). This operation is designed to straighten my stump which was beginning to bend inwards and therefore make limb fitting more difficult. I didn't mind spending time in hospital as there is plenty to keep you occupied (don't seem too well or they will have you doing school work!) but I hated spending long periods of time on crutches and not being able to do the things I love to do.

I have done lots of different sports, I have skied, mountain biked, played football, skateboarded, played rugby and even managed the miserable 3 mile cross country races I remember around the school playing fields (I never came last). However, I particularly enjoy water sports: water skiing, kayaking, sailing, water polo, swimming and surfing. I started swimming lessons when I was 4 and I enjoy nothing better than being in the water. When I was 14 I joined the Dragons swimming club in Caerphilly in South Wales and began weekly training. After training with Dragons for a few months I qualified for the Welsh disability swimming squad and in a year my coach (Alan Iles) suggested that with a lot of hard work I could qualify for the Welsh Commonwealth Disability Squad. I was amazed at the progress I had made. It took months of preparation and weekends of training away in 50m pools but I made the team through grit and determination and without my studies suffering. I swam in the 50m freestyle heats and came 4th in my heat but 13th out of 21 overall and I was the youngest swimmer. My goal 7 years ago was to swim in the paralympics and another commonwealth games by the time I was 20, unfortunately my studies took a forefront as I had begun the long slog to becoming a Vet. I still swam competitively and I missed the paralympic team by a whisker but at University, swimming as much as I needed to became harder and student life became more important. I played waterpolo for the university and Birkenhead teams but my sporting career has stopped there for the minute.

It was a real honour to represent Wales and I may do something similar again one day, swimming the channel or kayaking around the coast of Britain perhaps!

I passed out of Liverpool University with the degree in veterinary science in June of this year and I started my first job in Newcastle Emlyn in West Wales in September. I am a mixed vet and in my average day I may see anything from a cat to an owl but my main interests are in cattle, sheep and horses. I fish for salmon and sea trout and go surfing in the summer evenings and just the other day I did a 10 mile walk up Cader Idris, the long way round! Whatever you want to do, whatever your ability, there is something out there for all of us to excel at and I am a great believer that having bigger hurdles for us to overcome gives us great strength.
It’s a waiting game

When Alexander Wise was born with only three toes, it was his mum Charlotte who diagnosed fibular hemimelia after researching the condition on the internet. Leg lengthening was recommended; read her personal experience as the family wait for the treatment to begin.

At my 20 week scan there was a small concern over one of his kidneys so I had a further scan at about 32 weeks and we even paid for a 4d scan to make sure all was well. Nothing regarding his leg bones was noticed on any scan.

On August 9th 2006 we welcomed Alexander Jacob (Xander) into the world. The midwife checked him over but I knew something was wrong as I caught a glimpse of his foot and my stomach jumped and I asked what was wrong with his foot. The midwife asked what I meant, I remember repeating and repeating “what’s wrong with his foot?” Finally she unwrapped him and said “oh he’s only got three toes” handed him to his dad as I wouldn’t take him, that horrifies me now but I was so shocked. The midwife didn’t say another word, disappeared and returned a couple of hours later!! We sat in silence, I kept crying and felt numb. Then the questions started, Was it my fault? Was it because I’d had a couple of glasses of wine whilst I was pregnant? Had I not eaten properly. Then the big one, would he walk? His right leg was noticeably shorter than his left, his foot was small and he only had 3 toes.

The maternity didn’t have a resident paediatrician so it would be the next day before we saw anyone. I was taken to the ward, I drew the curtains, I didn’t want to speak to anyone or look at their babies or have them look at mine. Paul had to leave to pick up our other son Joshua so I then spent the whole night crying. As for Xander he was great he slept well and was oblivious to the whole thing. The next day I was like a different person and turned into protective mum mode. He was my baby, we’d made him and we’d deal with whatever was going to happen. The paediatrician finally arrived looked at his legs and foot and ‘helpfully’ said, he’ll need a raised shoe and then left! I couldn’t wait to get home and get on the internet. We were discharged with no information, no offer of help or advice, just to expect an outpatients appointment in the post.

I think we had our first outpatient appointment about 8-10 weeks later. From what I remember we didn’t get much advice. Xander eventually had x-rays, hip tests and measurements taken. After several outpatient appointments we were moved to Birmingham Childrens’ Hospital under Mr Bradish. Xander had more X-rays and measurements taken. Mr Bradish thought the predicted leg difference at full growth would be around 7cm. He said limb lengthening would be a good option as Xander’s knee and ankle are fairly stable. The thought of this procedure terrified me but after further research it looked successful and Mr Bradish was the best man for the job.

The problem now became the waiting. It would be a year between appointments as nothing could be done as Xander is too small. We were also given orthotics appointments. The orthotics man we see is fantastic, he knew I was a paranoid mother who didn’t want huge horrible shoes for my son! Xander had started walking barefoot around his first birthday but was very unsteady. He had his first raised shoes and he hated them. There was a two size difference between each foot and they looked heavy and clumsy. He didn’t wear them very often and managed barefoot around the house.

At 16 months his second pair arrived, his dad put them on him and he walked unaided, brilliantly. We’d thought when he was born he may never walk but at 16 months old he did, not only walked but he ran, just as well as any other toddler. We were so happy, I can’t remember now but I’m sure I cried (again).
Throughout the following year he got stronger and faster and developed as well as he should. His speech developed rapidly, whilst we waited for our next appointment. The shoes got better as we started sourcing our own shoes and sent them away to be raised.

We met Mr Bradish again in 2008, with the bad news that he was leaving Birmingham for Great Ormond Street. He said we were welcome to carry on seeing him but by the time Xander needs his operation he will have most likely retired. I was gutted. I really wanted him to do the limb lengthening procedure. After a long year wait we are due at Birmingham Childrens’ Hospital in a few weeks, hopefully we will get a new consultant and more importantly some information on where we go from here. Xander is nearly 3 now and to be honest we don’t feel like we’ve got any where (medically speaking) it’s such a long waiting game.

STEPS has been so helpful and I’ve made a lot of cyber friends who offer great advice and much needed support. I’ve set up a hemimelia support facebook page which continues to grow. I’m so keen to raise awareness for these very rare lower limb deficiencies as I know what it’s like to feel alone and like it’s only happening to your child.

I’ve been continuously fundraising for STEPS since Xander turned one. As we spend so much time waiting I thought I’d use the time productively. Our family and friends have been brilliant and supported the fundraising events. My brother and his friend are running the Great North Run later this year for STEPS. It’s nearly 3 years since I spent night after night crying and worrying that my baby might not walk. The three years have gone so quickly, Xander has been at private nursery since he was 9 months and loves it. He takes part in everything, even sponsored bounces and toddles. He’s due to start school nursery in September, complete with tiny uniform. He can’t wait to go to school with his big brother. I’m sure the change will bring with it new problems and challenges particularly as he gets older and wants to keep up with the other children but I know he’ll be fine, he’s such a pleasant, cheerful and very funny little boy that doesn’t let anything get in his way. He’s football mad and loves the outdoors. He recently had his first pair of trainers that flash which he is thrilled with.

Although it can be tough and very emotional at times, especially with “where are my other toes mummy” questions, we know we’ve got a long road ahead with the limb lengthening etc. but he’s our little man and we will do everything we can to make his life as normal and happy as possible.

It’s a waiting game (continued)