

FACT SHEET

Congenital Limb Differences

Introduction

A congenital limb difference means that a baby is born with part or all of a limb missing. It occurs because the limb doesn't form completely during pregnancy.

A congenital limb difference can sometimes be identified when you have your pregnancy scans, but other times it is not discovered until after a baby is born. If you have just learned that your child has a limb difference you might be feeling scared, worried or overwhelmed. This is normal. Your child will find their own way of completing and participating in everyday activities, as well as doing some extraordinary things as well.

This fact sheet describes some common congenital limb differences. Medical terminology for limb difference has changed in recent years, so throughout this fact sheet we are using the most up-to-date medical terms alongside older ones that you may still come across on the internet. As these are brief descriptions, and all limb differences are unique to each child, it is important to discuss your child's individual situation with your healthcare team.

Body parts and limb difference terminology

You may hear healthcare providers use a variety of words to describe parts of your child's body or their specific limb difference. You may hear that your child's congenital limb difference:

- Affects their digits or rays (fingers or toes)
- Affects their upper limb (arm/hand) and/or lower limb (leg/foot)
- Are unilateral (affects one side) or bilateral (affects two sides)
- Are longitudinal, meaning a long bone or part of a long bone is missing
- Are transverse, meaning that a limb does not develop beyond a certain level and will appear as if it has been taken off.

The Limbs 4 Kids website provides brief definitions of a wide range of words related to limb difference - limbs4kids.org.au/about-limb-difference

Treatment of limb differences

Limb differences are unique to each child. The limb difference can range from mild to severe and may come with other health conditions. For this reason, there is no 'one size fits all' solution, and a customised plan to address your child's specific needs must be made with you.

A multidisciplinary team is needed to collaborate and develop your child's management plan. Within the team you may have paediatric doctors (rehab physicians and surgeons), physiotherapists, occupational therapists, social workers, prosthetists (artificial limb makers) and a variety of other health professionals involved. It all depends on your child's needs and functional goals.

To help children develop the best way of achieving their goals, they may benefit from assistive devices (orthoses) or an artificial limb (prosthesis). Some children may have surgery to enhance their abilities and others may simply require education and guidance. The goal is to help children develop independence and live meaningful lives.

Causes of congenital limb differences

The development of a child's limbs occurs very early in pregnancy, even before some people realise they are pregnant.

There are many different causes of congenital limb differences:

- Genetic - it can be related to a family's medical history, but this is not always the case.
- Environmental exposure - such as being exposed to certain medications.
- Blood flow during limb development.
- Amniotic band sequence.
- As part of a syndrome, where limb difference is not the only part of the body affected.
- It is also not uncommon for there to be no identified cause for the congenital limb difference.

Amniotic band sequence

Amniotic band sequence is the name given to a collection of differences caused when stringy bands in the fluid that surrounds the unborn baby get entangled around various body parts. The type of limb difference a child is born with will depend on which part of the body the amniotic bands have affected.

Commonly this involves a limb and/or fingers and toes, but the face and body wall may also be affected. A strand wrapped around fingers or toes can cause them to join together (syndactyly). In other cases, the strands wrap around a limb or digit (ray) so tightly that it restricts blood flow and stops it from forming.

Why these bands form in the first place is not known, though there are two theories. It may be due to a problem in their initial formation or a result of early breaking apart of the sac that surrounds the baby in the womb. These bands can sometimes be seen during pregnancy scans, but not always.

Common congenital limb differences - lower limbs

Lower congenital limb differences are ones that affect legs, feet and/or toes.

In the leg there is a thigh bone (femur), which connects to the lower leg bones of the shin. There are two lower leg bones - the larger inner bone, the tibia, and the outer thinner bone, the fibula. The foot and ankle are made up of a number of bones and are organised into five toes (rays).

Longitudinal Deficiency of Fibula (Fibular Hemimelia)

Most cases of longitudinal deficiency of the fibula occur sporadically (that is, there is no identified cause).

A child born with fibular hemimelia has a fibula bone that is partially or completely missing. The child's leg will look shorter than the unaffected leg, which is referred to as a 'leg length discrepancy'. This leg shortening is commonly associated with other changes in the same leg.

The tibia bone may appear shortened and bowed, the femur shortened; the foot may be smaller and bent at the ankle with limited movement. In addition, there may be toes (rays) missing and/or the knee joint may be unstable. Many children with fibular hemimelia will have only one leg affected (unilateral), although it can affect both legs (bilateral).

Longitudinal Deficiency of the Tibia (Tibial Hemimelia)

Most cases of longitudinal deficiency of the tibia occur sporadically, though some are associated with other abnormalities of the body (such extra or missing fingers, shortened femur, missing kneecap, deafness, cleft palate, changes in the spine) and several syndromes.

A child born with tibial hemimelia has a tibia bone that is missing or misshapen. The child's leg will look shorter than the unaffected leg, which is called 'leg length discrepancy'. The child's foot is often normal but may also have fewer toes (rays) and bent in an unusual position. The child's knee may also be unstable or unable to fully straighten. Most children with tibial hemimelia will have only one leg affected (unilateral), although it can affect both legs (bilateral).

Proximal Femoral Focal Deficiency (PFFD)

Most cases of PFFD are thought to occur sporadically or for no known reason, though some are inherited (passed down through the family).

In a child born with PFFD the upper part of the femur (thigh bone) is shortened to various degrees. The child's

leg will look shorter than the unaffected leg, which is called 'leg length discrepancy'. In more severe cases, the foot of the affected leg approaches the same level of the opposite knee.

Often the hip joint will not be well developed, the femur will be pulled upwards and turned outward (rotated), and the knee joint unstable. Most children with PFFD will have only one leg affected (unilateral), although it can affect both legs (bilateral).

Common congenital limb differences - upper limbs

Upper limb congenital limb differences are ones that affect arms, hands and/or fingers.

The arm has one upper bone near the shoulder, called the humerus. There are two bones in the forearm, the radius, which is on the thumb side, and the ulna, which is on the little finger side. The wrist and hand bones, like the foot, are arranged in rays (fingers).

Transverse Forearm Deficiency (Below-Elbow Limb Difference)

Transverse forearm deficiency is often referred to as a 'below-elbow limb difference'. Most cases are thought to occur as a result of a lack of blood supply to the developing limb.

A child born with this type of limb difference has an arm that has stopped forming below the elbow. This means the child is born with a part of their forearm missing, and without a hand and fingers, though sometimes underdeveloped fingers, called nubbins, are present at the end of the stump. As this type of limb difference has a similar appearance to that of an amputation of the arm, it is sometimes referred to as 'congenital amputation'.

Children born with a transverse forearm deficiency can also have a shortened humerus and an unstable elbow joint on the same side.

Longitudinal Deficiency of the Radius (Radial Hemimelia/Radial Club Hand)

Most cases of longitudinal deficiency of the radius are thought to occur sporadically, though some are inherited, and up to a third of cases are associated with a more complex medical syndrome which can involve blood, heart and kidney abnormalities.

A child born with longitudinal deficiency of the radius presents with shortening or complete absence of the radius bone, variable involvement of the thumb, and

forearm shortening and curving. The curve created in the forearm, wrist and fingers may cause restricted finger movement and weaker grip strength.

This type of limb difference may affect both arms, but the severity is different on each side.

Longitudinal Deficiency of the Ulnar (Ulnar Hemimelia/Ulnar Club Hand)

Most cases of longitudinal deficiency of the ulnar are thought to occur sporadically, or for no known reason.

Children are born with the ulnar bone missing or shorter than normal and may have abnormalities of the elbow and forearm. The hand and wrist are almost always affected and up to 90% of children with this limb difference have missing digits, which will impact on their hand function.

Most children with longitudinal deficiency of the ulnar will have only one arm affected (unilateral).

Common congenital limb differences - upper and lower limbs

Some limb differences can occur in both upper and lower limbs.

Syndactyly

A child with syndactyly is born with fingers or toes (rays) that are fused or "webbed" together. Syndactyly occurs because the fingers or toes fail to separate, and thereby remain webbed. Most often it involves the space between the middle and ring fingers, and occurs spontaneously, though it can be inherited (run in families) or less commonly, occur as part of a syndrome. Both hands and feet may be involved.

There are different types and degrees of syndactyly:

- Incomplete syndactyly means the webbing doesn't extend all the way up to the top of the fingers or toes.
- Complete syndactyly means that the webbing extends all the way up to the top of the fingers or toes.
- Simple syndactyly means that the fingers or toes are joined only by soft tissue.
- Complex syndactyly means that adjacent fingers or toes are joined by bone or bony cartilage, as well as soft tissue (rare in children).
- Complicated syndactyly means abnormally shaped, extra or missing bones are involved in the fusion.

Treatment is required if the webbing is interfering with a child's hand function or dexterity or the ability to wear certain footwear. This may become evident with growth and the rapid development of skills. The nature of surgery will depend on the type of syndactyly and the fingers/toes involved.

Symbrachydactyly

A child with symbrachydactyly is born with fingers or toes (rays) that have not formed properly. They may be webbed or fused together, short, stiff or missing. It differs from other hand conditions in that the underlying structure of the hand or foot (bones, muscles, nerves, ligaments) is commonly affected. The cause of symbrachydactyly is not known and thought to be due to a disruption of the blood supply to the developing limb early on in pregnancy.

There is large variation in the severity and number of digits affected. In severe cases there are no fingers/toes or a small big toe/thumb. In some cases, the missing fingers or toes are replaced with small fleshy stumps (nubbins). Symbrachydactyly can be treated with surgery but not all cases require treatment. Surgery is offered to improve hand function or for cosmetic reasons. Occupational therapists can help children make best use of the affected hand.

Most children with symbrachydactyly will have only one limb affected (unilateral). It is very rare to affect feet and hands at the same time.

Ectrodactyly

A child with ectrodactyly is missing their central (or middle) fingers or toes, leaving a "V" or "U" shaped cleft or gap which divides the hand or foot into two parts. For this reason, ectrodactyly is often referred to as 'split hand/split foot malformation' (SHFM) or 'cleft hand/foot'. When a cleft is present, the ectrodactyly often affects both hands (or feet), and in these cases ectrodactyly is usually inherited or passed down through the family. Ectrodactyly may look very different between family members.

In other cases, there may only be a fifth digit and no cleft, and these are usually the result of a new gene mutation. There are also syndromes associated with this condition, and a form of SHFH is associated with

deafness and therefore the child's hearing should be checked.

Treatment options vary depending on the severity and type of deficiency. Modification to shoes may assist some children. Reconstructive surgery can optimise hand function or be done for cosmetic reasons, if required.

Polydactyly

A child with polydactyly has extra fingers or toes, which may range from a small bump to a fully developed finger or toe. Often the extra finger is next to the thumb/big toe or little finger/little toe, and may be connected by skin, muscle or bone. Extra digits are common and often run in families, though occasionally they occur as part of a syndrome. Polydactyly usually affects one hand or foot rather than both.

Polydactyly is usually treated in early childhood with the removal of the extra finger or toe, if it does not move well and gets in the way (such as a foot with an extra digit that cannot fit into a shoe) or for cosmetic reasons.

Accessing support

If you have been told that your baby has a congenital limb difference, please remember you are not alone. Your healthcare team are there to support you in understanding what this means and the treatment available.

Limbs 4 Life can also assist by connecting you to other families who also care for children with limb differences. The Limbs 4 Kids website also has a wide range of information, personal stories and publications to assist you in your journey.

Limbs 4 Life would like to acknowledge the support of the Queensland Paediatric Limb Difference Clinic Team in developing this fact sheet.

www.limbs4kids.org.au | T: 1300 782 231 | E: kids@limbs4life.org.au

The Limbs 4 Kids program is an initiative of Limbs 4 Life, Australia's peak organisation for amputees and those living with limb difference. Limbs 4 Kids ensures that all Australian children and young people with limb difference, as well as all of those who care for them, receive access to information and support.