

Information for families and new parents

Congenital Hand Difference

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Introduction

Currently, less than 50% of limb difference can be picked up at the 20 week scan. All parents tell us that they experience shock, guilt and in some cases, sorrow at the time of the surprising news that their baby has a limb difference. This is a totally normal reaction; you need to give yourself time to process your emotions.

If you have found out at your 20 week scan that your baby has a limb difference, the information can allow you time to come to terms with things, time for you to research, and be prepared for the birth of your baby.

In the beginning you need to take the time to come to a level of acceptance, take time to heal and understand. By taking this time and giving yourself space you will feel so much stronger and bolder about how your amazing journey with your Reach child will unfold. Reach children are incredible, their resilience and ability to adapt is amazing.

A recent survey of 261 parents about their early experience of receiving the surprising news that their baby has an upper limb difference confirmed the following:

- Relevant information was only provided to 28% of parents
- Approximately 50% of parents saw a specialist within 3 months
- Over 90% of parents searched for information themselves
- Over 70% of parents felt unsupported in the early stages after receiving the surprising news

The underlying cause for a congenital upper limb difference was discussed with only 37.5% of parents.

It is important to remember that the majority of children with congenital hand differences are otherwise healthy.

We hope that this booklet will help improve your understanding of upper limb differences and what you might expect on the journey with your child.

Mental health

Many parents find mental health support beneficial when they find their baby has an upper limb difference. This can come in many forms including counselling, Cognitive Behavioural Therapy and Psychology.

You can access referrals to mental health support through your Health Visitor, GP or any other health care professional.





Information for families

“Congenital” means that a condition was present at birth.

This information booklet is for new parents of babies with hand differences to help with the understanding of ‘how’ and ‘why’ these occur and to offer a simple overview of some of the common congenital hand conditions and their possible treatment.

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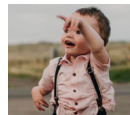
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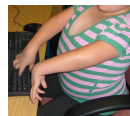
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Causes of Congenital Hand Differences (CHDs)

What are CHDs?

'Congenital' means 'from birth'. Congenital hand differences (CHDs) occur when a child is born with a hand that had formed differently from what we would expect of a 'normal' hand. This happens in up to 1 in 2,000 live births. Please note that we prefer the term 'differences' rather than 'abnormalities' because that is exactly what it means, i.e., it is just 'different' rather than 'abnormal'.



The lines show how long I was at

4¹/₂ weeks



5¹/₂ weeks



6 weeks



7 weeks

How do CHDs happen?

Embryology is the study of how a foetus develops. This image shows how the human embryo grows inside the womb. Note that the hand is fully formed within 4 to 8 weeks of a woman becoming pregnant, and the disturbance of growth occurs earlier at a point where she does not even know that she is pregnant.

The developing embryo uses a network of signals which work together to form the hand. This happens in three directions to produce the 3D shape of the fully developed human hand.

One of the most important regulators of this development is a protein called **sonic hedgehog (SHH)**. Sometimes, there is either too much or too little SHH, which causes the hand to develop differently.

Why has this happened?

Depending on when and where in the growth pathway these signals differ, development of the upper limb and hand will be affected in different ways.

It is not yet clear why CHDs happen. They may be due to genes – which 'plan' how the foetus develops in the womb – or the environment in which the baby grows inside the womb, or the external environment.

However, we know that most CHDs happen for unknown reasons and are not related to the genes or behaviours of parents. **It is important to remember that parents are not to blame when CHDs happen.**



There are several support charities for children and families affected by CHDs which we encourage you to consider:

Reach offers families the opportunity to connect and share stories with other families affected by upper limb differences. Through their website and family weekend events, this charity provides useful information for families as a child reaches each milestone:

www.reach.org.uk

Details of other organisations which can support you and your child are available on the Reach website.

Remember if you are struggling in any way or just want some advice, please ring the Reach Head Office on 0300 365 0078 or email us at reach@reach.org.uk. The Reach family is an enormous community and support network and full of parents, carers, young adults and children who have a wealth of knowledge and experience. We can always put you in touch with a Health Professional or a parent/family in a similar situation, remember you are not alone.



Treatment

How are CHDs managed by the health service?

- Hand differences are rare, and each hand difference is unique.
- Not all hand differences actually need any treatment. A personalised management plan will need to be made by discussing your child's hand difference when you see a Hand Surgeon who specialises in treating children's hand differences.
- Most hand differences do not need immediate treatment – the exception being arthrogryposis and some conditions with wrist and elbow involvement, when early intervention with therapy including stretching and splints can be helpful.
- Diagnostic information from x-rays may not be helpful until the child is older because the bones are very soft in the first few months after birth.
- Surgery, if appropriate, will aim to improve function by improving the way the hand and arm works. Consideration is also given to the appearance/cosmesis of the hand.
- Occasionally the hand difference in your child might be part of a 'syndrome' which includes other 'abnormalities'. Your clinician may therefore offer to refer you and your child to a geneticist especially if you have a family history of limb differences. However the majority of children with congenital hand differences are otherwise healthy.

Hand therapy:

Hand therapy is the non-surgical management of hand disorders and injuries using physical methods such as exercise, splinting and wound care.

- The hand therapist can assist with emotional and psychological support, as well as with restoration of hand function.
- Hand therapy has a crucial role in the recovery from hand surgical operations. Hand therapy is integrated into the hand surgery team.

Surgery:

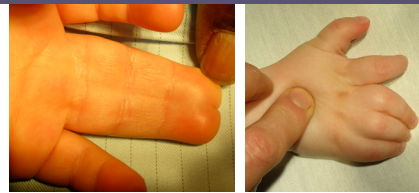
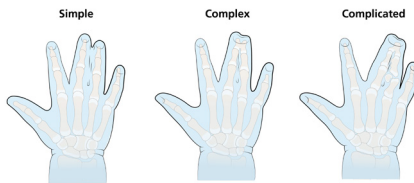
The aim of all surgical procedures is to improve function and cosmesis. It is not possible for surgery to restore a 'normal' hand.

- The best time for a surgical procedure is often between 12 and 18 months of age when the child is old enough to safely undergo an elective procedure. At this age recovery from surgery is relatively quickly achieved.
- A protective bandage will be worn after the surgery until the wounds are healed. A child of this age is less distressed when a bandage prevents movement. Removing a bandage is also relatively stress free in this age group.
- However sometimes it is better to wait until a fuller pattern of use of the hand is seen before reconstructive surgery is offered.

There is no right or wrong decision with regards to surgery! Children will adapt extremely well and will become independent adults who contribute to society with or without a surgical intervention.



Syndactyly (Webbed Fingers)



What is Syndactyly?

Syndactyly is a common condition in which a child's fingers or toes do not fully separate during development, causing "webbed" spaces. The spaces between two or more fingers or toes may be webbed. Sometimes it is only the skin that is joined, but in other cases the fingers can also share tendons, nerves, blood vessels and bone.

What causes Syndactyly?

During the early weeks of pregnancy, the child's fingers and toes form in a "mitten" of skin. By the end of the second month of pregnancy, the extra skin dissolves, and the fingers and toes should separate. Syndactyly occurs when the fingers and toes do not fully separate during development.

Some forms of syndactyly are inherited, while others are sporadic, meaning the condition can occur even if it doesn't run in the family. Most cases of syndactyly are isolated and occur in an otherwise healthy child.

How Is Syndactyly treated?

Your child's surgeon will help you determine the appropriate treatment option. Some forms of mild syndactyly may not need treatment. If a reconstructive operation is necessary, web spaces are created carefully to minimize scarring. Sometimes, a small piece of skin from another area of the body, called a skin graft, will be needed to help cover the space between the fingers after they are separated.

Surgery is typically performed between 12 and 18 months of age.

After surgery, the child will wear a protective cast and sling to allow the hand to rest and heal. Follow-up visits will be scheduled to monitor the healing and function of the child's hand.

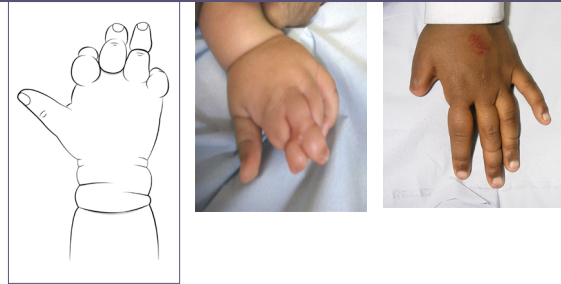


Post op Syndactyly separation



Conditions

Amniotic Band Syndrome/Constriction rings Syndrome/Sequence



What is 'ABS'?

Amniotic Band Syndrome is also known as Constriction Ring Syndrome and it happens when fibrous bands of the amniotic sac (the lining of the uterus that contains a fetus) gets tangled around the developing baby. Most children with amniotic band syndrome will have bands on more than one part of the body.

What causes ABS?

There may be some vascular blood supply upset which occurs very early in the pregnancy which causes some loss of fingers or hands or feet and leads to a brief slowing down of the growth of the baby's limb.

How is ABS treated?

Treatment is different for each child. Some shallow bands may not interfere with function and may not require surgery. If your child has a deeper amniotic band, they may need one or more operations to help to improve both the function and the appearance of the hand. Sometimes, if the band around a limb is very tight, the operation to release this may need to be done at around 6 weeks of age.

However, most of the other effects of ABS such as syndactyly (joined fingers) and the shallow bands around the digits can be treated surgically at around 18 months of age.



Sybrachydactyly (Short or Missing Fingers)



What is Sybrachydactyly?

Sybrachydactyly describes small or missing fingers or a missing hand. There may also be webbed fingers or a short hand or forearm.

Short or missing fingers occur in many forms, most commonly because the fingers did not develop completely during very early pregnancy. The condition is usually present on one side only, with a possibility of muscle abnormalities of the chest wall on that side as well. (See: Poland's Syndrome).

What causes Sybrachydactyly?

The exact cause of the condition is unknown, and it does not seem to run in families. It is thought that there is insufficient developing tissue for all of the parts of a full arm and hand to form.

How is Sybrachydactyly treated?

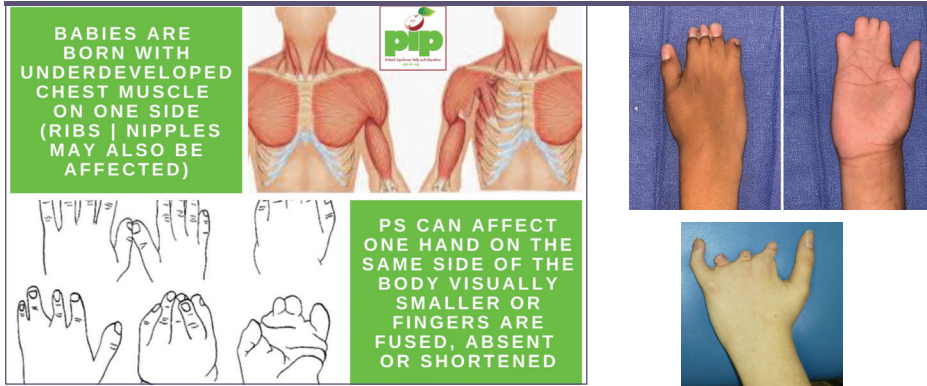
Your doctor will discuss with you the best treatment options for you or your child. While there is no way to restore the hand completely, treatment options are available, including surgery to deepen the webs between the fingers and to stabilize the fingers with bone grafts. However, surgery is often not needed.

The goal of treatment is to allow the child to be as independent and confident as possible in using his or her hand in daily activities.



Conditions

Poland's Syndrome



What is Polands Syndrome?

Poland syndrome is rare, affecting 1 in 20,000 live births. It is present at birth (congenital), and consists of underdevelopment of some or all of the chest, shoulder and upper limb on one side of the body. The hand difference, if present, is a form of Symbrachydactyly. It affects males more frequently than females.

What causes Polands Syndrome?

The cause of Poland syndrome is thought to be due to a temporary alteration in blood flow in the developing shoulder girdle and upper limb during pregnancy at approximately seven weeks. Individuals affected by Poland syndrome will lead a normal and healthy life.

How is Polands Syndrome treated?

Each case is assessed individually. Hand difficulties, as well as underdevelopment of the chest and breast can be reconstructed, usually by plastic surgeons.

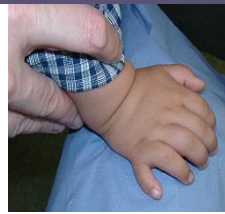
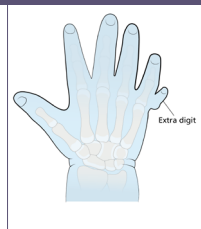
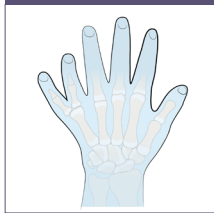
More information can be obtained from the Poland Syndrome Charity:

www.pip-uk.org





Polydactyly/Extra fingers/Digits



simple syndactyly



complex polysyndactyly

What is Polydactyly?

Polydactyly is one of the most common hand conditions present at birth. The condition can appear as any form of an extra digit, anywhere from a small, raised area to a complete digit. The affected digit may also appear "split."

What causes Polydactyly?

In some cases, polydactyly is genetic, but it can also accompany other hand conditions, such as syndactyly (webbed fingers.) It is then called:

Complex Polysyndactyly

This is where there is a combination of extra digits which are joined together. It can be associated with other syndromes which can be passed down in families.

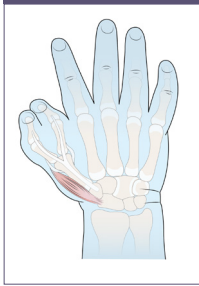
How is Polydactyly treated?

Treatment for polydactyly varies in complexity. A minor procedure under local anaesthetic shortly after birth, can remove small extra digits or those with a narrow base. However, when the base is broad or if the extra digit is rooted deeply in the hand, reconstructive surgery may be the best option when the child is old enough to safely undergo an elective procedure. Operating on the bone, joint, ligaments or tendons may be necessary to create the most functional hand possible for the child.



Conditions

Extra thumbs/Thumb Duplication/Thumb Polydactyly



Example of before and after surgery for duplicated thumb

What is Thumb Polydactyly?

Thumb polydactyly is when more than one thumb is present on a hand. There are many appearances and varieties of thumb duplications, varying from a broad fingernail to a complete double thumb. Polydactyly can involve bone, joint, muscle, tendon, nerve, blood vessels and/or skin. Usually it occurs on one hand only, and neither thumb is completely formed.

What causes Thumb Polydactyly?

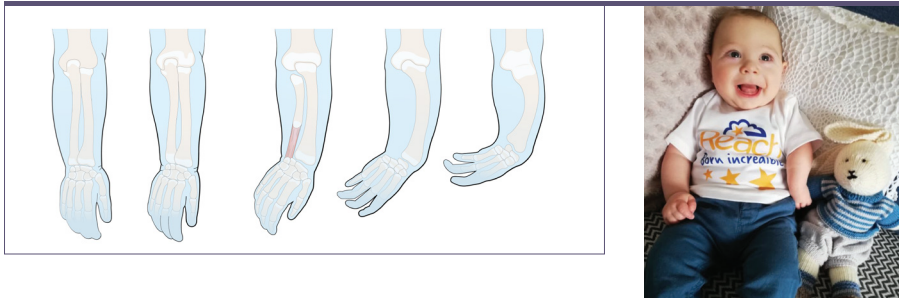
Thumb polydactyly can occur in many forms, most commonly because the child's thumb formed into two parts during the second month of pregnancy.

How is Thumb Polydactyly treated?

Treatment options vary based on the anatomy of each thumb but generally involve reconstructive surgery of the bone, ligaments and tendons. Sometimes, parts of the two thumbs may be combined to build the best possible thumb for the child.



Ulna Longitudinal Growth Disturbance



Ulna Longitudinal Growth Disturbance

is known by other names as follows:

- Ulna Longitudinal Deficiency
- Ulna Hemimelia
- Ulna Club Hand
- Ulna Dysplasia/Aplasia

What is Ulna Longitudinal Growth Disturbance?

Children with Ulna Longitudinal Growth Disturbance are born with their wrist in a bent position toward the little finger side of the hand. The fingers and thumb may also be affected. Other muscles and nerves in the hand may be unbalanced or missing.

There are two bones in your baby's forearm: the ulna, on the little finger side of the arm, and the radius, on the thumb side. Ulna Longitudinal Growth Disturbance happens when the ulna and other soft tissues of the hand do not fully develop.

What causes Ulna Longitudinal Growth Disturbance

Most of the time, the cause of Ulna Longitudinal Growth Disturbance is unknown. It may sometimes run in families, particularly as part of an inherited syndrome, like ulnar mammary syndrome and Klippel Feil syndrome. It can also be associated with syndromes that are not inherited, like Cornelia de Lange syndrome.

How is Ulna Longitudinal Growth Disturbance treated?

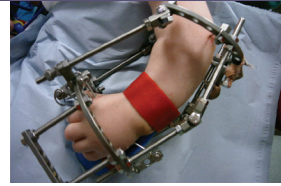
Each child will be unique and will require a discussion with a Children's Hand Specialist to work out an individualised management plan.

Surgery and Non-surgical therapy will aim to rotate the limb into a better position, straighten the wrist if necessary and address the fingers and thumbs in order to achieve at least 2 digits which can be in a position to grip an object.



Conditions

Radial Longitudinal Growth Disturbance



Radial Longitudinal Growth Disturbance

is known by other names as follows:

- Radial Longitudinal Deficiency
- Radial Hemimelia
- Radial Club Hand
- Radial Dysplasia/Aplasia

Radial Longitudinal Growth Disturbance is a rare condition that affects the forearm, and can affect one or both arms.

There are two bones in your baby's forearm: the ulna, on the outer side of the arm, and the radius, on the inner side. Radial Longitudinal Growth Disturbance occurs when the radius does not form properly. This causes the wrist to bend toward the thumb side of the forearm.

Radial Longitudinal Growth Disturbance also affects the soft tissues and flesh of the forearm. The arrangement of muscles and nerves may be unbalanced, and some muscles and nerves may be missing. The thumb may be absent or smaller than usual with poor joints and missing muscles and/or tendons.

What causes Radial Longitudinal Growth Disturbance?

Radial Longitudinal Growth Disturbance usually occurs by chance, although for some people it can also run in the family. Doctors and scientists do not know why some children are born with this condition.

There are no known links between this condition and the parent's lifestyle or anything the mother may do during pregnancy.

Radial Longitudinal Growth Disturbance can be associated with several congenital syndromes, including those affecting the heart, digestive system, and kidneys.

How is Radial Longitudinal Growth Disturbance treated?

All children will adapt to their own situation remarkably well as they grow and develop their manual skills. They will find solutions to most tasks expected of them. The aim of any surgery is to make those tasks easier to achieve.

There are various options for treatment and the specialist doctor will explain which are most suitable for your child. This decision about suitable treatments is influenced by the severity of your child's Radial Longitudinal Growth Disturbance and their general health including other symptoms. It also depends on you and your family's feelings about treatment, as some options involve a number of operations and quite involved care at home, which will require determination from all involved.

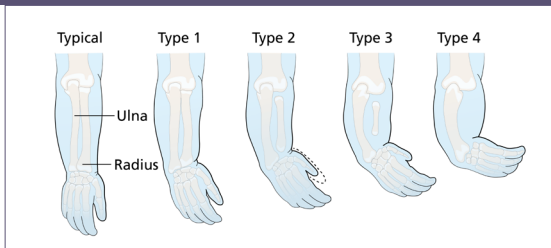
It is not possible surgically to restore a normal hand and forearm.

However, it is possible to make the forearm straighter, and longer and to realign the wrist.

Non-surgical hand therapy and splints

Non-surgical treatment with stretches and splints is usually required in all types of Radial Longitudinal Growth Disturbance.





In mild cases, early in infancy, you will be shown a series of stretching exercises to carry out on your child's wrist with the aim of increasing the range of movement.

In more severe types of Radial Longitudinal Growth Disturbance, stretches and splints are used. These are used before surgery to stretch the soft tissues. Splinting is also used after surgery to help maintain the corrected wrist position.

Surgery

A series of operations is sometimes recommended to those children with type II, III and IV radial club hand, where a straightening of the wrist may be considered helpful for functional and cosmetic reasons. Surgery tends to take place in phases. There are also some operations that are best done when a child is at a certain age. Your doctor may suggest the following operations:

Distracting the soft tissues of the wrist with an external fixator

This aims to correct the position of the wrist and is normally planned for when your child is walking, usually at around two years of age. There are two parts to this procedure. The first part involves surgery to attach an external fixator (a metal bar or frame) to your child's forearm and hand bones. After the operation, the soft tissues are gradually stretched – this process is called distraction. The two pieces of the external fixator are gradually moved apart to straighten the wrist, usually by a millimetre or so each day. Once the distraction phase has finished, the fixator needs to remain in place for several more weeks while the bone grows

stronger and the tissues remain stretched. During this time, you will be expected to help clean where the pins come through the skin and to perform the distraction. You will be supported and trained in how to do this and you will need to be seen regularly in the hospital to check that all is going as planned. The process of distraction is lengthy and demands commitment from you and your child. The usual time that the distractor is on the arm is between eight and twelve weeks.

Re-positioning the hand

This is an operation to remove the fixator and maintain the new position of the hand sitting on the end of the ulna bone. This will either be achieved by a tendon transfer for a 'radialisation' operation where movement is preserved, or a tendon transfer and bone fixation for a 'centralisation' operation, where most of the wrist movement is lost, but the wrist is fixed in a better position for function.

The bending of the wrist can recur as the child grows older and may require further surgery. For this reason, all children require follow up until adulthood when growth has finished, and the growth of the arm and hand are stable.

Forearm lengthening

Most children with Radial Longitudinal Growth disturbance have short forearms in the limbs affected. There are operations that can be used to lengthen the forearm but these are complex and rarely produce forearms of the same length. If surgery is possible, this would be offered during mid to late adolescence.



Conditions

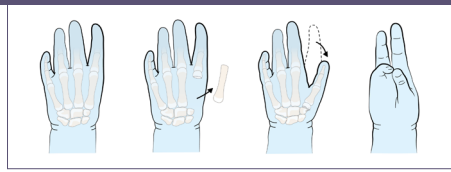
Thumb Hypoplasia and Aplasia (absent)



Pouce-Flotant/Floating Thumb



Post Op Pollicisation



Index Pollicisation Procedure

What is Thumb Hypoplasia and Aplasia?

Thumb Hypoplasia means that your child's thumb is unusually small or underdeveloped. Additionally, the bones and the muscles that move the thumb are involved. This condition is also commonly called hypoplastic thumb.

Thumb Aplasia means that your child's thumb is missing altogether.

What causes a child's thumb to be underdeveloped or missing?

The cause of this condition is unknown. However, it often happens together with Radial Longitudinal Growth Disturbance (see above). It can also happen in association with the following syndromes:

- a) Holt-Oram syndrome
- b) Fanconi syndrome
- c) VACTERL sequence.

How are Thumb Hypoplasia and Aplasia treated?

The decision about whether or not Thumb Hypoplasia or Aplasia should be treated is not always straightforward. Children adapt and can function with a missing thumb. If untreated, children who have no use of a thumb can learn to pinch objects using their long and index fingers. But they may have problems with pinch strength as well as activities that require holding larger objects. Surgery for Thumb Hypoplasia is designed to help with these functional issues and with appearance.

Thumb reconstruction surgery

This surgery reconstructs the ligaments in the thumb to stabilize the middle joint and improve function and stability by transferring a tendon from another part of the hand. It may also involve an operation to release the tight web space between the thumb and index finger using a skin graft.

Thumb reconstruction can be done when your child is around 18 months of age or between 3 and 5 years old.

Index Pollicization surgery

This surgery may be an option if your child has no thumb or severe hypoplasia. The operation involves creating a functional thumb by transferring another finger (usually the index finger) to the thumb position. Very often, the child will be using the index and middle finger to grip an object and the operation aims to improve this function by shortening and rotating the index finger to make it more like a thumb.

If there are no other pressing medical concerns that need to be addressed, pollicization surgery is generally performed when your child is between 6 to 18 months old.

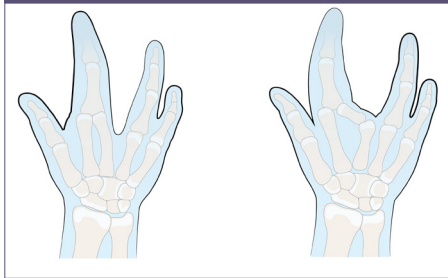
Occupational therapy

If your child has a mild case of Thumb Hypoplasia and their thumb is slightly short or weak, or if the web space between their thumb and index finger is slightly tight and prevents mobility, occupational therapy will help them adapt. If your child has surgery, occupational therapy will be essential after the surgery to maximize results.





Cleft Hand



What is Cleft Hand?

Cleft Hand is a rare condition in which the centre of a child's hand is missing a finger or fingers. Cleft Hand makes up less than 5 percent of all congenital hand differences.

Cleft Hand occurs when a child's hand doesn't develop fully during pregnancy. The condition can only sometimes be detected on a routine prenatal ultrasound. After the baby is born, the difference is visible.

Although Cleft Hand usually affects both hands, it can also occur in only one hand. Children born with Cleft Hand may also have cleft foot.

Not all Cleft Hands look the same. Typically, the centre portion of the hand is missing a finger or fingers and there's a V-shaped cleft in that space. Less often, clefts occur on the thumb side or the little finger side of the hand.

In the majority of children with Cleft Hand, the condition is an isolated occurrence that affects only the hands. Your child's doctor will check for other associated differences or syndromes, including cleft foot, cleft lip, and cleft palate.

What causes Cleft Hand?

Cleft Hand develops during pregnancy when the bones of the hand are forming. The exact cause is unknown, but scientists and doctors are learning more and more about the possible genetic causes (passed from parent to child).

How is Cleft Hand treated?

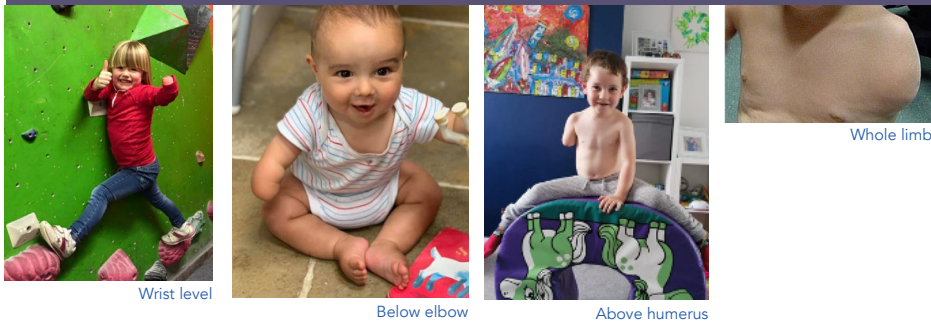
Not all Cleft Hands need treatment. If there are at least 2 digits which can work opposite each other, then a very good function can be achieved without any intervention.

Surgery needs to be individually determined and aimed at improving existing function and cosmesis by watching how the child uses his/her hand.



Conditions

Transverse Arrest/Amputations



What is a Congenital Amputation/ Transverse Arrest of the upper limb?

This happens when the limb doesn't develop fully during the very early stages of its development. The limb bud can stop growing at various levels – shoulder, above elbow, below elbow or wrist level.

It is usually seen in isolation and affects one limb only. No familial incidence has been shown.

Sometimes tiny digits/'nubbins' are seen at the end of the amputation 'stump'.

What causes Congenital Amputation/ Transverse Arrest?

It is thought that something affects a few cells in the growing end of the limb bud at a very early stage of the development of the baby, before the mother even knows she is pregnant.

How is Congenital Amputation treated?

Most children adapt well. The child will use the gap between the arm and the body and/or the elbow and/or both hands together to hold objects. The skin of the arm and little digits, if present do have feeling which is very useful for the child.

Prosthetic limbs are available in many different forms and the technology is advancing very fast

- Purely cosmetic
- Myoelectric
- Mechanical/3D printed
- Task orientated - e.g. Koala mitt

The NHS/HSE rehabilitation service will offer advice on the use of these at any stage of your child's journey

Surgery may occasionally be necessary if the end of the amputation 'stump/nubbins' become inflamed.

Babies and children do not usually feel any pain in their amputation stump but it may feel cold and change colour at times. Simply keep the whole child warm.





Arthrogryposis (Limited Movement and Stiff Joints)



What is Arthrogryposis?

Arthrogryposis, meaning stiff joints, is a condition that includes multiple joint contractures and lack of muscle development. The most commonly affected joints are in the child's hands, wrists, elbows, shoulders, hips, knees and feet.

What causes Arthrogryposis?

The cause of arthrogryposis cannot be identified in most cases. When joints are not moved for a period of time before birth, extra connective tissue develops, and it can result in joint contractures. In this case, the joint is fixed in a stiff or locked position and the tendons connecting the joint do not stretch to their normal length, making joint movement difficult.

- Limited joint movement can be the result of one of the following:
- Muscles that do not develop properly
- Muscle diseases
- A fever or virus during pregnancy that damages cells transmitting nerve impulses

- A decreased amount of amniotic fluid
- Failure of the central nervous system and spinal cord to form correctly
- Tendons, bones, joints or joint linings that develop abnormally

A genetic cause has been recognized in about 30 percent of cases.

How is Arthrogryposis treated?

Physical and occupational therapy, as well as splinting and exercising, may help improve flexibility, position of the child's joints and use of the upper arms. Surgery may be necessary to release joint contractures that do not respond to therapy.

The goal of both operative and nonoperative treatment is to help children gain independence in daily activities and to prepare them for school and adult life.

Further information can be found at The Arthrogryposis group Website:

www.arthrogryposis.co.uk/index




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