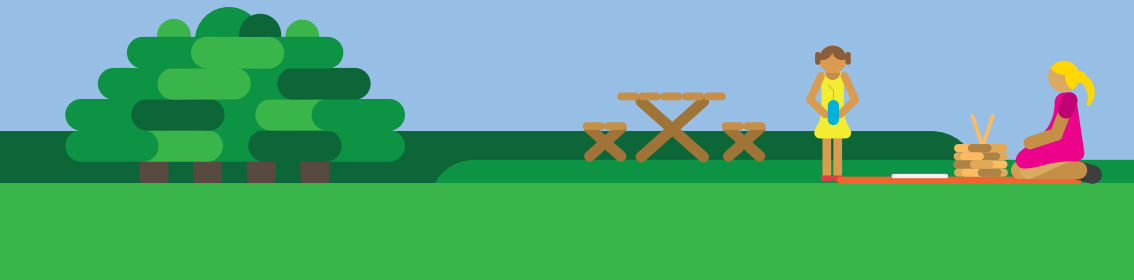
An introduction to CDH

This information booklet has been produced by CDH UK, a registered charity offering support to families affected by CDH. The following information is to provide the reader with an insight into CDH from diagnosis to birth.



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Disclaimer: Whilst every effort has been made to ensure the accuracy of the information contained within this booklet, it is not intended to replace or substitute the views or information provided by the person responsible for the medical care of any individual.

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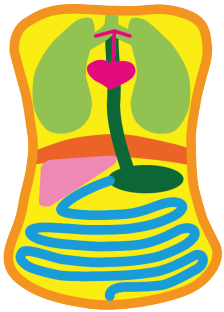
It's highly likely that you've never heard of CDH before. So here is a detailed description of what it actually is.

CDH is the abbreviated name for Congenital Diaphragmatic Hernia. 'Congenital' means born with , 'Diaphragmatic' means of or affecting the diaphragm; which is a thin sheet of muscle that helps us to breath and keeps our chest and stomach contents separate. 'Hernia' is a general term used to describe a 'bulge' or 'protrusion' of an organ, for example the stomach, through the structure or muscle that usually contains it. CDH occurs in approximately 1 in 2500 births and accounts for around 8% of all major congenital abnormalities.

There is no firm evidence that it occurs more in one particular sex. However, some studies have suggested that males are more commonly affected than females with a ratio of 3:2. Familial clusters (running in families) have been observed in less than 2% of cases and the risk in future pregnancies is said to be 2%. There is currently no known cause or risk factor, but studies have suggested that it is probably multifactorial, meaning there maybe a number of factors involved including environmental and genetic.
The diaphragm normally develops in your unborn baby by around the 7-10 week gestation period of your baby's development and CDH occurs when the diaphragm fails to form correctly or fails to develop in your unborn baby, allowing the abdominal contents to herniate into the chest cavity, which in turn prevents the lungs from developing properly. Depending upon at what stage the abdominal contents (also referred to as 'Viscera') herniated, how much, and the size of the hole in the Diaphragm will determine how much your unborn baby's lungs and other internal organs, such as the heart, have been affected.

'CDH is the abbreviation for Congenital Diaphragmatic Hernia'

> The most common form is left-sided and accounts for around 85% of all cases



A correctly formed abdomen

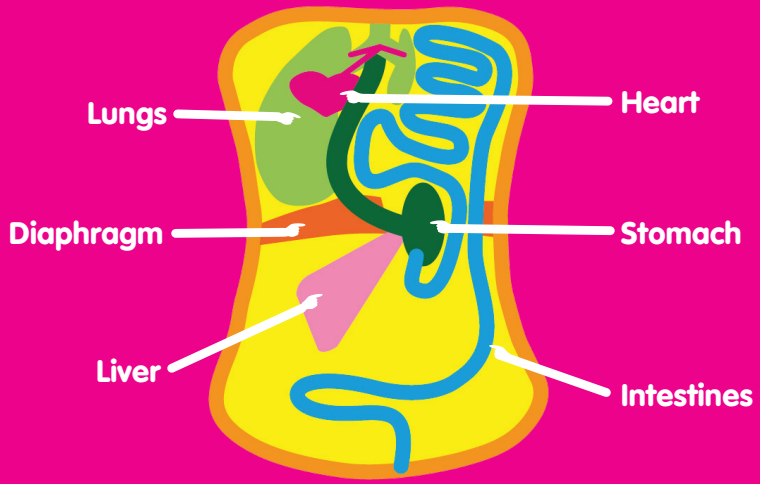
There are different types of CDH; the most common type is Bochdalek which accounts for over 90% of diaphragmatic hernias and is usually on the left side. Morgagni hernias are less common and are found behind the sternum (breastbone) with most being slightly to the right side. Diaphragmatic eventration occurs when the diaphragm is still intact but is weak and abnormally high in the chest (can be either unilateral or bi-lateral). They may simply be refered to as left-sided, right-sided or bi-lateral.

In a left-sided hernia, varying amounts of abdominal contents can herniate, including small and large bowel, stomach, spleen and sometimes the liver. In right-sided hernias, it is usually only the large bowel and or liver that herniates. Bilateral hernias are uncommon and as the term suggests; affects both sides. In most cases and particularly in left-sided hernias, the condition is isolated, which means that there are no other associated anomalies. Rarely, CDH is part of a chromosomal syndrome such as Edward's Syndrome (Trisomy 18), Patau's syndrome (Trisomy 13), Down's Syndrome (Trisomy 21) and Turner Syndrome (Monosomy X). However, a Karyotype test is usually offered to confirm that the hernia is isolated. Uncommonly, other syndromes such as Cornelia De Lange and Fryn's syndrome have also been associated with CDH.

Other features of CDH can include variable degrees of Lung Hypoplasia (incomplete

development of the lungs), Pulmonary Hypertension (increased pressure in the pulmonary arteries) and heart defects. These are often difficult to diagnose and confirm to what extent they are present until after your baby is born. Other malformations can occur and are principally the cardiovascular (heart, lungs etc) genitourinary (kidneys, bladder etc) and gastrointestinal systems (stomach, bowel etc).

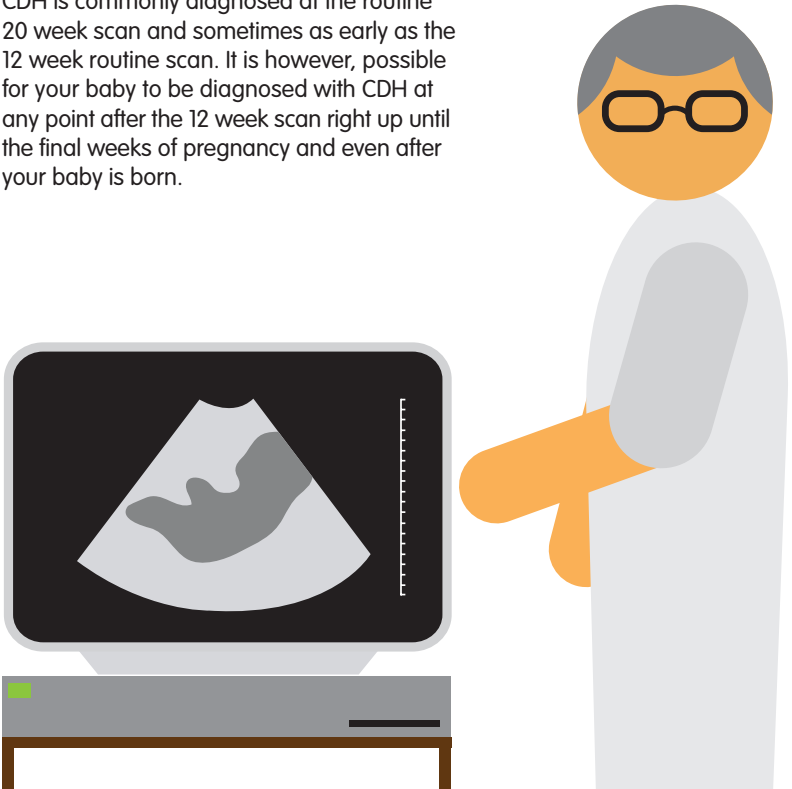
Currently the prognosis (outlook) for babies diagnosed with CDH remains statistically at 50%, however, some hospitals claim to have a higher survival rate. A measurement called LHR (lung:head ratio) is often used by Doctors as an indicator to help them to assess the severity of the hernia and to assist them with planning the best management for your baby. This is taken during an ultrasound scan and your Doctor can explain this further to you. As each case is different, each case warrants its own outlook; some babies with extremely good prospects before birth fail to do well and some babies with very little lung and a poor outlook will do extremely well.



An example of a CDH abdomen

Diagnosing CDH during your pregnancy.

CDH is commonly diagnosed at the routine 20 week scan and sometimes as early as the 12 week routine scan. It is however, possible for your baby to be diagnosed with CDH at any point after the 12 week scan right up until the final weeks of pregnancy and even after your baby is born.



It is picked up by the person carrying out the scan (sonographer) when it appears organs are not where they should normally be, or there is something unusual about the scan. You will then be invited to attend another scan session to have the diagnosis confirmed.

Below are some of the signs that may be picked up on a scan:

- Abdominal contents (viscera) are noticed in the chest cavity where the lungs are positioned, or if the lung/lungs appear abnormal in size, shape or position.
- When there is difficulty picking up an image of your baby's heart.
- If the heart is pushed over to the left or right (mediastinal shift).
- Polyhydramnios, which is an increase of amniotic fluid surrounding your baby.

CDH can sometimes be associated with a chromosomal disorder (although this is rare), such as Edwards Syndrome, Fryn's syndrome and Cornelia-de-Lange syndrome to name a few. This would be confirmed by carrying out a Karyotype test, which is done by testing the amniotic fluid surrounding your baby. The procedure to obtain a sample of amniotic fluid for this test is called an amniocentesis.

Occasionally CDH is not picked up during routine scans and is diagnosed shortly after birth.

Extremely rarely CDH is diagnosed later in life during routine medical check ups or procedures, or following further herniation of abdominal contents.

The risk of recurrence in future pregnancies is extremely low and is given as a 2% increased risk. You can ask to be referred to a Genetic counsellor to discuss this small increased risk.

Expect extra support and care throughout your pregnancy.

Throughout your pregnancy you can expect to be more closely monitored. Some of the things you can expect are:

- Extra ante-natal appointments to closely monitor the pregnancy and your baby. This may involve a referral to a local Fetal Medicine unit.

- MRI (Magnetic Resonance Imaging) to attempt to obtain the size and volume of your baby's lungs, which may provide the Doctors with additional information to prepare for your baby's arrival.

- Amniocentesis to check for chromosomal abnormalities, because rarely CDH is associated with other syndromes. It is entirely your choice whether you opt for this test, and if you feel you do not want to have this procedure carried out discuss this with your Doctor.

- A referral to a tertiary centre, which is a specialist centre within a hospital that has the experience to look after your baby during your pregnancy and after he or she is born. Whilst your local hospital will try to ensure that the hospital they refer you to is as close to where you live as possible, it is not always the case and can sometimes mean you may have long distances to travel.

- The opportunity to meet with the medical team who will be caring for you and your baby during and after the birth. This may include a neonatologist (newborn baby doctor), Paediatrician (doctor who specialises in caring for babies and children), a paediatric Surgeon (a doctor who specialises in carrying out surgery on babies and children), a consultant Radiologist (a Doctor specialising in x-raying, scanning and MRI scanning) and a Fetal medicine specialist (a doctor who specialises in caring for unborn babies and pregnant women).

- You may also be given the opportunity to have a look around the neo-natal intensive care unit (NICU) so that you know what to expect when your baby arrives.

- If the centre you are referred to has trained counsellors, you may be offered a counselling service. If in doubt ask the Doctor responsible for managing your care.

Please be aware that not all of the above may apply to your individual care plan.

Your pregnancy should progress normally from a physical and general health point of view. You may however, develop Polyhydramnios during the pregnancy, which is an increase in amniotic fluid surrounding your baby. This will be monitored by the person carrying out your scans and the person responsible for looking after you throughout your pregnancy, such as a consultant obstetrician or Fetal medicine specialist. If the fluid increases too much and you feel uncomfortable, or the doctor feels it necessary, the excess fluid will be drained from around your baby. This is carried out by local anaesthetic and whilst it may be uncomfortable, it should not be painful and your Doctor will explain the procedure fully in advance.

An experimental procedure called FETO (Fetal Endoscopic Trachael Occlusion) may be offered in some cases of CDH. This involves the placement of a balloon into your baby's trachea to attempt to improve the development of the lungs whilst your baby is growing in the womb. FETO is classed as a minimally invasive procedure carried out by a specialist medical team, but is not without risk. The procedure is currently only carried out at one UK hospital for severe CDH cases and there is no firm evidence to show that this is successful in reducing mortality rates in infants, however, it is shown to have had some success in certain cases. There is a certain criteria required for this procedure and your Doctor can explain what this is.

Creating the right birthing plan for you.

It is normal for the birth to be planned in advance, so you may be given a set date for the birth of your baby, this can be anything from 37 week gestation onwards and your doctor should discuss this with you before you approach the end of your pregnancy. This usually involves inducing the birth to ensure your baby arrives on the date planned and at the Tertiary Centre.

This ensures that all of the medical team involved are present at the birth and that there is an available cot in NICU for when your baby arrives. Do not be surprised if the planned date is changed, as sometimes other emergencies in the department arise or cots are unavailable.

If everything is going well with you and your baby the Doctor will probably arrange the induction for as near to 40 weeks (full term) as possible.

During pregnancy, a member of the medical team will discuss with you what is going to happen when your baby is born and will encourage you to ask as many questions as you need.

You can be expected to deliver your baby normally and be offered pain relief choices, however, if things are not progressing or if your baby is distressed you may require a caesarean section (C-section). Both you and your baby will be monitored extremely closely during your labour and because of this you may find that you are unable to move around and are restricted as to your birthing options.

‘Your labour will be planned to the greatest detail to make sure all the medical and support team are present at birth’

You can discuss this with your Doctor or Midwife, who will tell you exactly what you can expect and what your choices are.

Occasionally, things don’t go according to plan and your baby arrives sooner than expected. In this situation you will normally be admitted to your local hospital, who will then arrange for the appropriate plan of action. They should usually have been informed of your case by the tertiary centre, so they’ll know exactly what to expect.

After your baby is born, he or she will be transported by a specialist neo-natal team by ambulance to a tertiary centre with an available cot. Depending on how you are after the birth you should be able to go with your baby to the hospital.

What happens after your baby is born?

When your baby arrives you may notice that he or she may not cry and that they are blue in skin colour (Cyanotic), do not be alarmed by this, even babies without CDH may not cry or have a bluish colouring (Cyanosis). Some Doctors prefer for babies not to cry, so that they are not using their underdeveloped lungs, which reduces the risk of damage to them. Likewise do not be surprised if they cry, this is also normal.

As soon as your baby is delivered, you can expect a lot of activity in the delivery room. The priority is for the Doctors to attend to your baby immediately and usually this involves intubating your baby, which is inserting a flexible tube into your baby's trachea (airway) via your baby's mouth, the insertion of an NG tube (naso-gastric tube)

> **The care immediately after birth is in both your interest and your baby's interest**

into your baby's stomach, to release any air, and administering medication to keep your baby still and sedated.

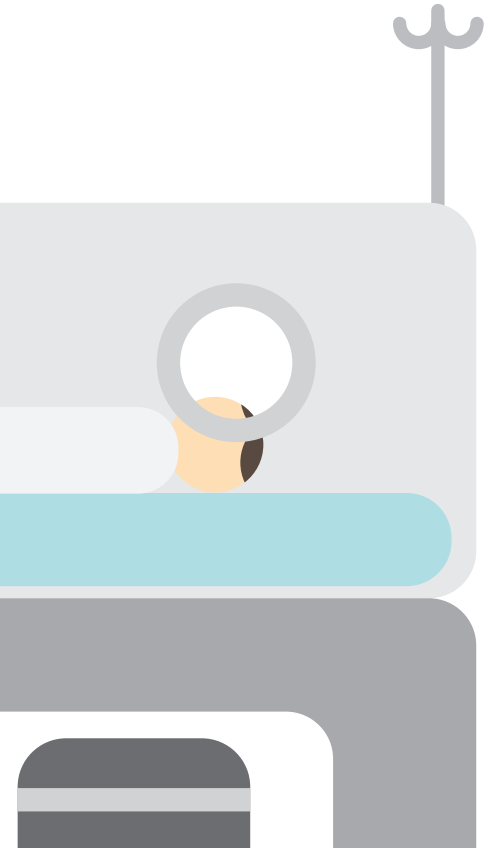
This is all carried out to enable artificial ventilation (life support). This will be carried out as quickly as possible in order to stabilise your baby. In addition to this your baby's apgar score will be taken and recorded, this is a series of observations undertaken at the birth to establish the condition of your baby and is carried out on all newborns. It is usually carried out at 1 minute after birth and 7 minutes after birth and offers useful information to the Doctors.

Sometimes your baby may require resuscitation immediately after birth if they are not breathing at all.

You will probably be unable to hold your baby after birth, but once he or she is stabilised in the delivery room you may be offered a quick glance before they take your baby to NICU. It may then be a few hours before you can see your baby, because he or she has to be put onto the chosen ventilation equipment and will have to undergo a stabilisation period, x-rays, blood tests and other monitoring procedures.

Also, if you have encountered a difficult birth or had a C-section it may not be possible for you to be up and about until the Doctor's are satisfied that you are well enough. This can be distressing, but be assured that this is in both your interest and your baby's interest and as soon as it is possible you will be allowed to see your baby.

Taking care of your baby over the coming days.



The first 24 hours in NICU can be a critical time for your baby. Your baby will be placed on a ventilator and will be constantly monitored by the doctors and nurses on the unit. There will be a lot of noisy equipment around as well as other sick babies and this can be distressing. The Doctors and nurses will explain what the equipment is doing for your baby and how your baby is doing.

Depending on your baby's needs, the ventilation method could be conventional or HFOV (High Frequency Oscilation Ventilation). Sometimes your baby can stabilise quickly, but also his or her condition can deteriorate just as quickly.

‘The Doctors will explain what the equipment is doing for your baby and how they are doing’

Some babies who are placed initially on conventional ventilation may be transferred to HFOV, which is a more ‘gentle’ form of ventilation and is therefore less likely to damage the residual lung (existing good lung). Your baby may also receive inhaled Nitric Oxide (INO) which assists in the treatment of respiratory failure.

A rescue therapy called ECMO, (Extracorporeal Membrane Oxygenation) may be discussed if your baby is failing on ventilation, which is basically a lung, heart bypass machine. This allows your baby’s lungs to rest whilst supporting your baby’s life. There are only a few of these machines available in the UK and there is a certain criteria required before your baby can be considered for this treatment, as it is not suitable in all cases.

There is no current medical evidence to support that the use of ECMO is beneficial as a standard treatment to babies with CDH and most centres use the ‘Gentle Ventilation’ method.

Repairing the hernia.

Once your baby is fully stabilised and the Doctors are happy with your baby's blood gases, oxygen levels and general progress, your baby will undergo surgery to repair the hernia (there is no set timescale for this). The paediatric surgeon will probably have already explained prior to your baby's arrival, of the procedure that will be undertaken, but will explain this again to you prior to the operation.

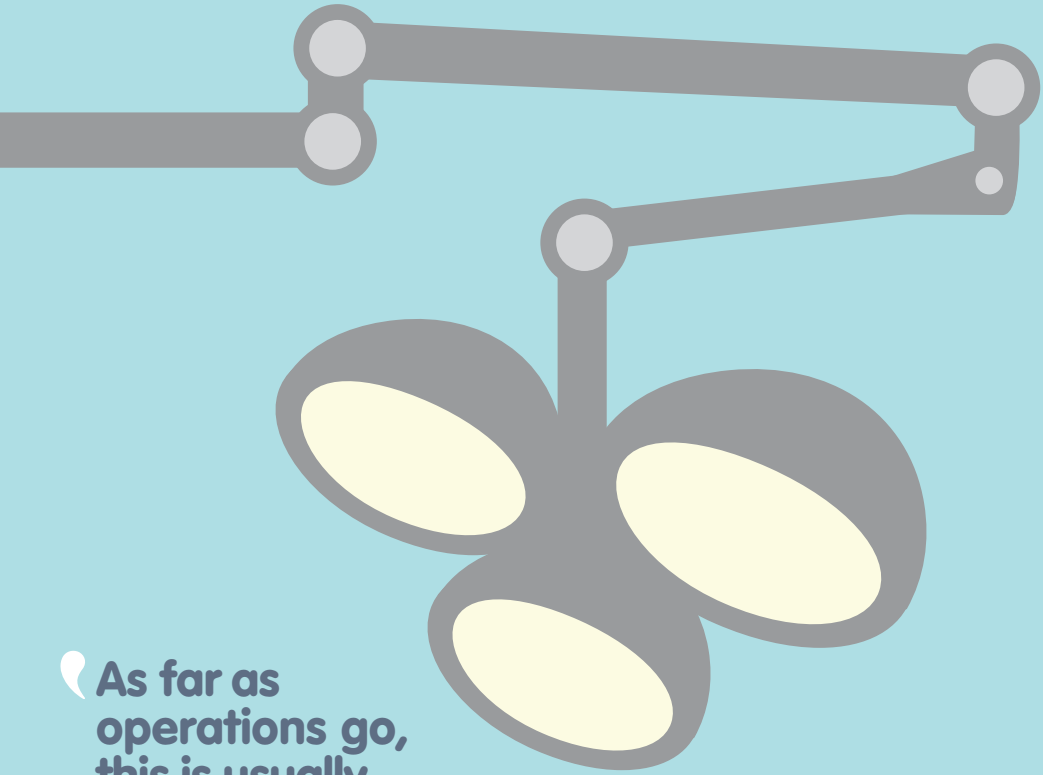
As far as operations go, this is usually a relatively straightforward procedure, whereby the surgeon makes an incision in your baby's abdomen on the side of the hernia, moves the abdominal contents back to where they should be and repairs the hole in the diaphragm.

Depending upon the size of the hole, the surgeon will decide whether to suture (stitch) the hole up or to attach a patch to repair the hole. The patch is usually made from a material called 'Gore-tex®'. He will also be able to ensure that there is no damage to the intestines and correct any problems he finds at the same time.

You will be fully informed after the operation of how it went and if there were any complications. Your baby will be returned to NICU and be closely monitored whilst recovering from the operation. It is not uncommon for babies to deteriorate after the operation, so be prepared for an emotional time.

If your baby recovers well and his or her condition remains stable, the Doctor's may decide to move baby from NICU to a surgical unit.

The scar left by the operation will fade as your baby grows and in most cases will be only a faint scar in the future.



‘As far as operations go, this is usually a relatively straightforward procedure’

Losing your baby.

Sadly, some babies lungs are just not compatible with life, which means that no matter how hard the Doctors try to stabilise your baby, all the signs indicate that your baby is extremely poorly and that his or her lungs just can't cope. This is often referred to as respiratory failure and is usually a result of severe lung Hypoplasia (under-development of the lungs).

Sometimes, there are other factors such as heart failure, deterioration during the operation or other complications.

The Doctors and nurses will explain why your baby is deteriorating and will discuss with you what course of treatment can be carried out to attempt to try and improve your baby's condition and what options are available. Unfortunately, there can come a time when the Doctors feel that it is in your baby's best interest to cease treatment. This is never an easy decision for parents or medical professionals to undertake and you will be given as much time and support as you need to make your decision.

Some parents find that their baby lets them know when the time arrives, as the parents can see that their baby is struggling. At this point you may feel that you want all of your family and friends who haven't yet had the chance to meet your newborn to visit the hospital.

The NICU staff will try and fulfil all of your requests with regards to visitors if your baby deteriorates, but remember that most of these units have strict visitor policies and they have to consider the needs of the other babies and their parents, so it is not always possible to accommodate all of your visitors.

It is a good idea to take lots of pictures and videos of your baby and usually the nurses will do this too, as well as keep a little diary of your baby's day to day care for you to take home. You may also be offered a parents room on the unit, so that you can be close to your baby at all times. It can also be arranged for you to have your baby baptised on the unit and the nurses will be very supportive in helping you make these arrangements.

When the time comes to remove your baby from the ventilator, you can choose to be present or to wait for the nurses to bring your baby to you. This may be the first time you and your partner have actually held your baby and this alone can be extremely overwhelming. You are encouraged to bathe and dress your baby and to hold them and talk to them for as long as possible.

You may also wish to take some footprints or handprints of your baby and some photographs.

The hospital usually give you a 'memory box', containing things like your baby's blanket from their cot, heart monitor pads, diary and photographs and also useful information such as how to register your baby, support group information and funeral arrangements.

Everyone reacts differently in this situation and there is no right or wrong way of dealing with emotions or feelings. You may find it useful speaking to a hospital counsellor or hospital chaplain if they are available. Family and friends are also invaluable at this difficult time.

Leaving hospital and living with CDH.

The best possible outcome is if your baby continues to improve and is transferred to your local special care baby unit and eventually you are able to take your baby home. The length of time your baby will be in hospital can be anything from 6 weeks to over 12 months depending on whether your baby improves quickly or experiences complications.

Once your baby is home you can be expected to receive visits from outreach nurses and health visitors, who will help you with all aspects of your baby's ongoing recovery and issues. If all goes well your baby will have no lasting problems at all and will grow up to be a normal healthy adult. However, here are some of the things your baby may experience as a consequence of CDH:

- Feeding issues such as your baby being slow to breast or bottle feed which may affect growth and weight gain and in extreme cases; a failure to thrive. Occasionally a tube (NG tube) placed into your baby's nose and down into the stomach is necessary to administor feeds and sometimes medication.

- Reflux (gastric acid flowing from the stomach into the oesophagus), which can be distressing for your baby and is usually treated with medication. In severe cases a procedure called a nissen fundoplication is carried out where the surgeon improves the natural barrier between the stomach and the oesophagus. This prevents the flow of acids from the stomach into the oesophagus, and strengthens the valve between the two, which stops acid from backing up into the oesophagus as easily.

- Speech and developmental issues resulting from artificial ventilation and long periods of hospitalisation. Your child will normally be referred to a specialist and speech therapist to deal with these issues.

- Hearing loss can occur and nobody knows exactly why this happens. Again, your baby will be monitored throughout childhood by an audiologist (Hearing specialist) with regards to his or her hearing.

- Re-herniation is uncommon, but can be serious if it occurs. The signs that this may have happened are as follows:

  - your baby or child is generally unwell
  - loss of appetite,
  - abdominal discomfort and or pain
  - difficulty breathing,
  - vomiting (may contain bowel contents).

If you notice any of these signs contact your GP immediately or call an ambulance.

- On-going medical issues
  For further information on on-going problems or medical issues please email support@cdhuk.org.uk as we can provide further literature and information upon request.

Understanding your emotions and feelings.

There is no right way or wrong way to feel, we are all individuals who cope with situations and emotions in different ways. CDH can also have a huge impact on your family and friends. We have listed below some of the normal emotions that you, your family or even your friends may feel at some point

- Devastation
- Fear
- Anxiety
- Anger
- Numbness
- Impatience
- Negativity
- Uselessness
- Neglect
- Guilt

You may have other children who you feel are being neglected because of your time given to baby and this again is normal.

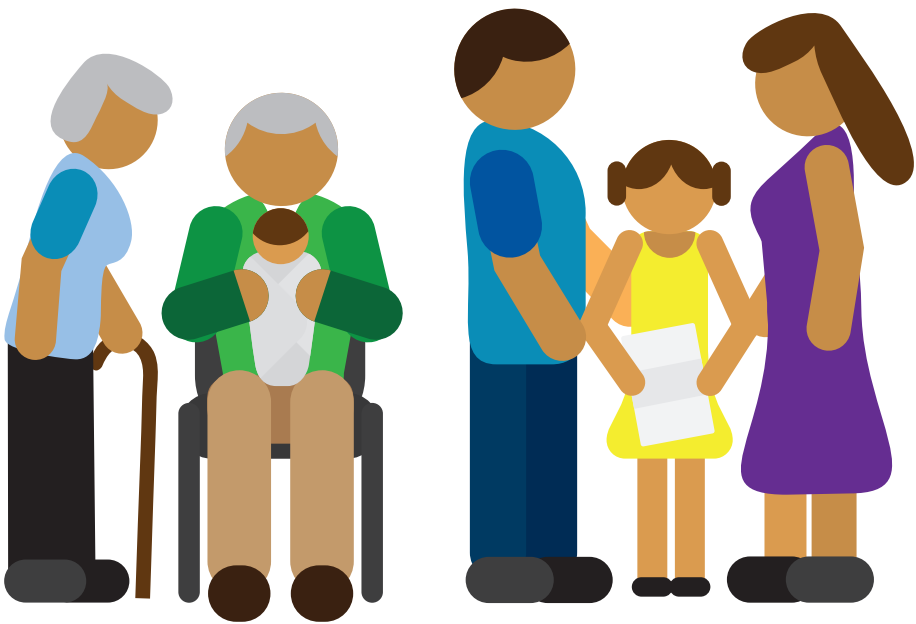
Having family and friends on hand who can look after them, take them to school and on outings will be helpful and will serve as a distraction until you are with them.

Talking about your baby and how your baby is doing to siblings, showing them pictures and videos of your baby and encouraging them to draw a picture or make something for the new arrival will help them to cope and understand what is happening.

CDH UK can offer you a range of support, from information and advice to simply lending an ear. We can find other parents nearby who have experienced the same situation as you and help with costs such as travelling and parking known as our 'Home to Hospital' scheme. We can also put you in touch with other organisations which can help (see useful contacts and websites).

Free professional bereavement counselling is also available by request (subject to conditions).

‘Never underestimate the strength of support that family, friends and other parents can provide’



If you have any questions write them down here ready for when you see your consultant.

Helpful Contacts & Links

ARC Antenatal results and choices www.arc-uk.org

CDH Australia www.cdh.org.au

Asthma UK www.asthma.org.uk

Scoliosis Association (UK) www.sauk.org.uk

Child Bereavement UK www.childbereavement.org.uk

Stillbirth And Neonatal Death Society (SANDS) www.uk-sands.org

CDH UK webshop www.spendandraise.com/cdhuk

Medical Research Articles www.pubmed.gov

Bliss - Baby Life Support Systems www.bliss.org.uk

Bounty - Support for new parents www.bounty.com

Birth Defects Foundation www.specialsource.org/condition_results.cfm?condition=2169

British Heart Foundation www.bhf.org.uk

CDH UK my charity page www.mycharitypage.com/cdhuk

Ebay for charity CDH page donations.ebay.co.uk/charity/charity.jsp?NP_ID=37468

Contact A Family (CAF) www.cafamily.org.uk

CDH UK is a member of Rare Disease UK www.raredisease.org.uk

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https://twitter.com/#!/CDHUK_CHARITY

We're CDH UK and we're here to give help, support and advice.

CDH UK helpline
0800 731 6991

Website
www.cdhuk.org.uk

Email
support@cdhuk.org.uk

Alternatively write to us
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King's Lynn
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cdhUK The Congenital Diaphragmatic Hernia Charity

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