

Supporting You Through Your Pregnancy

A Handbook for Parents
after a Prenatal Diagnosis

ARC

Antenatal Results & Choices ●

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This information has been written for parents who are continuing their pregnancy after the diagnosis of a chromosomal, structural or other condition in their baby. We hope that carers, family and friends of the parents will also find it helpful.

In this handbook 'you' applies to both parents when both are involved or to either one when that is relevant. For convenience, we use 'she' when referring to midwives.

Our aim is to help you gain the information, advice and support you may require in order that your needs as a parent are met as well as those of your baby. You may find some of the contents distressing or feel that certain sections are not relevant to your situation; this is because we want to include all parents, whatever has been diagnosed.

Prenatal diagnoses cover an extremely wide range. Some conditions can be treated; some cannot. Sadly, some parents may be faced with a diagnosis which means their baby's future after birth is very uncertain, or may know that their baby is going to die.

If you would like to talk about any of the issues raised in this handbook, please contact ARC on **020 7713 7486** or **info@arc-uk.org**



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DEALING WITH THE SHOCK

Being told that your baby has a condition or is not developing as expected will have an emotional impact on you. The news will most often come as a shock and you may face a mixture of confusing feelings. These may not necessarily be any less or different because your baby has been diagnosed with a condition that is not life threatening or not considered by health professionals or people around you as 'serious'.

The strength of your feelings may take you by surprise. But remember, like all parents, your hopes had centred around the arrival of a healthy baby and suddenly that has been taken away from you. To react to this situation in a very powerful way is normal.

You will need to allow yourself time to adjust to the news that this pregnancy is different to the one you had planned and hoped for. Many parents speak of needing to grieve for this lost dream before they are able to cope with the present reality. This is entirely normal and do allow yourselves the time and space to do this.

You may experience a wide range of conflicting feelings; again this is not unusual. Initially, you might find that the shock means you refuse to believe the diagnosis. Some parents feel a sense of guilt. It is natural to ask yourselves if your baby's condition could have been avoided. You may even start blaming yourselves, each other or believe that hospital staff were in some way responsible.

There is very rarely any kind of connection between something you think you did or didn't do and what has been diagnosed in your baby. Even if you know that what has happened is nobody's fault, you might find yourself feeling angry.

Some parents go through an intense feeling of failure; they may feel they have failed their baby, themselves and each other. Other parents talk about going through phases when they try to detach themselves emotionally from the baby or they have conflicting feelings about the pregnancy in general.

You may also feel isolated. It may seem that you are very alone and are experiencing something that no one else can understand. Please feel that you can call the ARC helpline and talk through your feelings with us whenever you need to. ARC may also be able to signpost you to organisations who can put you in touch with others who have shared similar circumstances.

Facing a prenatal diagnosis is almost always a stressful situation. There is no 'right' way to feel and we are all unique. Some parents worry that they are going 'mad' because of the strength or unpredictability of their reactions, when in fact, what they are going through are natural responses to distressing circumstances. As some of the emotions you will experience will be very painful and unlike anything you have encountered before, it can help to be able to talk them through. One of your team of health care professionals, particularly your midwife, may be able to spend time talking about you and how you are feeling. You could ask if there is a counsellor available at your hospital or GP's surgery. Unfortunately, there can sometimes be quite a long wait for an appointment. ARC may be able to advise you of counsellors/therapists in your area.

There may be special considerations if you are carrying twins. A condition may be diagnosed in one or both of your twins. You may have to face difficult and painful decisions because one twin is affected and the other is not. In some cases you may be offered a selective reduction of the pregnancy. You can call the ARC helpline to discuss your particular situation. It may also be useful to contact TAMBA, the Twins and Multiple Births Association, (<https://www.tamba.org.uk/>)

YOUR DECISION TO CONTINUE THE PREGNANCY

It may be that you are, and always have been, entirely committed to continuing the pregnancy, whatever the nature of your baby's condition. In some cases, the possibility of termination is not raised because the baby's condition can easily be treated. However, for some parents it may have been very difficult to make the decision about whether to continue or not with the pregnancy. It may seem that your health care professionals are being unhelpful because they cannot tell you what to do. They cannot, and indeed should not, try to push you in any direction. Only you know what is right for you, your family and your baby.

Sadly, there may be those around you who are not as supportive as you might expect. There may be pressure or criticism from those close to you. Some people, even those you regard as close, may make insensitive comments, which at best are not useful and at worst can be very hurtful.

Ultimately, you need to feel that you have made the choice that is right for you. Your decision will be respected and supported by all the health professionals responsible for your care and an appropriate plan for your pregnancy will be worked out with you.

AFTER THE INITIAL DIAGNOSIS

Being told that your baby has a particular condition often comes as a shock; and it is not always easy to take in information when you are distressed or anxious. However, it is important that you have all the information you need about what has been diagnosed in your baby. Support is helpful, so try to make sure your partner, a friend or family member is with you whenever you see a doctor. The person who goes with you can help make sure you go away having asked all the questions you intended. Always ask any health professional you see to explain things to you slowly and carefully and ask for a clear explanation of any complicated medical terms.

It can be useful to write down a list of questions to ask before any appointment, just in case you forget something when speaking to your consultant. There should be someone, perhaps a specialist midwife or genetic counsellor, who you can contact between appointments with any questions or concerns.

You may be referred to a number of health professionals who are specialists in different branches of medicine. As well as your obstetrician, who specialises in pregnancy and birth you will probably have appointments with a paediatrician. A paediatrician will talk to you about the care for your baby after birth. There is a glossary at the back of this booklet which lists other specialists you might meet.

Your appointments should take place in an environment that is as private and quiet as possible, although this can sometimes be difficult in a very busy hospital. The discussions may be distressing but your doctors will want to give you as accurate an assessment as they can about the outlook.

At any time during a consultation, you can ask to be left alone for a while. You can then use this time to think and talk about what you have heard or even just have some breathing space. If you feel that you cannot take in any more information, you can always request another appointment. Ask if there is information about the condition available, either online or printed, which you can read in your own time and at your own pace at home.

In many cases your health care team will be able to tell you a great deal about your baby's condition. However, some conditions are less common and little is known about them. In this situation, your specialists will do their best to find out as much as possible.

Unfortunately, there may be situations when health professionals cannot tell you exactly what is wrong with your baby or the implications of what they have found and this can be a cause of great uncertainty and anxiety for you.

Many parents and professionals use the Internet as a source of information. There are some excellent websites, but bear in mind that the quality and accuracy of information varies. You can contact us in the ARC office and we will suggest resources, if they are available.

The organisation Contact: for families with disabled children (<https://contact.org.uk/>) or Genetic Alliance UK (<https://www.geneticalliance.org.uk/>) may be able to put you in touch with support groups for parents who have had children with the condition that has been diagnosed in your baby.

In some cases the diagnosis may mean that your doctors cannot be entirely sure about what will happen during the pregnancy or once your baby is born. The outlook may change, sometimes significantly, at any time during the pregnancy. This lack of certainty can be very emotionally draining as you may find your emotions veering from hope and optimism to worry or fear. It can be comforting to have someone you are able to express these feelings to. This person may be a family member or friend, or perhaps someone from your local community, such as a faith leader. Remember you can also speak to someone at ARC whenever you want to.

WHAT YOU MIGHT NEED TO KNOW

You may need to ask more than one of your health professionals to get the answers to the following questions.

- Can you explain exactly what you have found in my baby?
- How certain is the diagnosis? If it is not certain, how and when will it be confirmed?
- What exactly does it mean for my baby?
- Will we need any special care or treatment before birth?
- What will happen at birth?
- Will my baby need treatment immediately after birth?
- Where will this treatment take place?
- Will my baby need to stay in hospital? For how long?
- What can you tell me about how my baby will develop before and after birth?
- What is the future outlook?
- Where can we get more information?
- Are there any support organisations that might be helpful?
- Is this a condition that may affect future pregnancies? If so can an appointment be made to see a genetic counsellor?

Who will be responsible for my care?

It is likely that you will be referred to a specialist fetal medicine unit so that your baby can be given expert care. You should be given the names and titles of all the health care professionals who will be responsible for your care. Depending on your baby's condition, these could include a consultant obstetrician, midwives, a paediatrician, and a neonatal or paediatric surgeon. There should be one person among your care team who can act as a 'key worker' for you; ideally this should be someone you find easy to communicate with and who you feel able to ask for help or support. A midwife can be valuable in this role as she will be able to help with all your concerns about the pregnancy, not just those surrounding your baby's condition.

If you are referred to a specialist fetal medicine unit within a teaching hospital, trainee doctors or research fellows could be present at some or all of your appointments. While they do have to learn, your permission to have them there should always be sought and if at any time their presence makes you uncomfortable, do not be afraid to ask your consultant to tell them to leave.

What kind of extra care will my baby and I need?

The level of extra care and monitoring needed during the pregnancy will depend on your baby's condition and sometimes your medical history. Make sure you understand what will happen and are clear about why particular monitoring or procedures are necessary and that you are happy for them to go ahead.

Do not feel that all your choices and control over the pregnancy are taken away simply because of the diagnosis; any kind of treatment or test should only go ahead after discussion with you.

When and where will the appointments be?

Depending on where you live, the specialist hospital you are referred to could be some distance away and you might find yourselves having to organise transport and make arrangements with work or for childcare. If you are going somewhere unfamiliar, contact the hospital beforehand to check on transport options, best routes and parking facilities if necessary.

Employers are legally obliged to allow mothers the paid time off they need for antenatal care. Most employers are very understanding; if yours is not, ask for a letter from one of your health care professionals stressing the need for your appointments. It may be more difficult for partners to negotiate the necessary time off. Many consultants are aware of the difficulties and will offer you appointment times when it is more convenient for both of you to attend. When you are allocated an appointment time that is problematic for you, do ask whether an alternative can be arranged.

Sometimes your specialist unit serves quite a wide area and can be quite busy. This may lead to long waiting times, so you might want to take something to read or to occupy yourself before your appointment. In this circumstance, it may not be easy to have young children with you. The unit should be able to give you some idea of how long you can expect to be there.

If you are on benefits, you may be entitled to help with travel costs for your hospital appointments. You can find out more here:

<https://www.nhs.uk/using-the-nhs/help-with-health-costs/healthcare-travel-costs-scheme-htcs/>

What about the mother's physical and emotional wellbeing?

You might find that much of your energy and concern as well as that of your team of health care

professionals is concentrated on your baby and his or her condition. Please do not disregard your own needs, both emotional and physical.

Sometimes the fact that you are a mother-to-be can be forgotten, and just because your baby has a condition does not mean that you will not have similar needs to other pregnant women. Your partner must also make sure their needs are addressed.

You may have particular concerns if you are first-time parents. Having a midwife as a 'key worker' for you throughout your pregnancy can be helpful for you as she will help make sure that the day-to-day management of your pregnancy is as smooth as possible.

You may have difficulties being around other expectant women and couples, e.g. at routine scans or antenatal classes. It is entirely natural to feel resentful towards or jealous of mothers-to-be who have 'sailed through' their pregnancies and have not had to face the upsetting news that you have. If your appointment times bring you into contact with pregnant women and you find this painful, tell your health care professionals so they can organise alternatives. You should be able to arrange to go for early or late appointments to avoid situations that are hard for you.

Taking part in antenatal classes may not feel appropriate for you. Some parents find it difficult to attend classes with parents who are not in the same situation. But if you would like to attend, you can ask what is available at your local hospital and you can talk to the person running the class so they there are aware and sensitive to your situation. You may want to see a programme for the classes and choose to go to the ones that you feel you would benefit from most. Attending the classes could be helpful in building relationships with community health professionals. Otherwise, you might be able to arrange for a community midwife to visit you at home.

There may be times when it may seem as if no-one is taking overall responsibility for your antenatal care. If your local hospital has referred you to a specialist unit, they might believe you are getting all you need there, while the specialist hospital will not be able to help with all the routine aspects of your care. You may prefer to visit the clinic at your local hospital for routine appointments. Make sure you make your wishes known. Ideally, everyone involved in your care will be aware of your situation, but communications between and within busy hospitals can sometimes fail. So you may find yourself having to explain your situation more than once to different health professionals. For some parents, this can add to their distress.

You may sometimes feel as if you are on an emotional rollercoaster and it can be helpful when you feel able to talk through your feelings. When this is not possible with one of your health care team, do call us on the ARC helpline. Please also be aware that the emotional impact of what has happened and is happening may hit you some time later and perhaps unexpectedly. Even if you are able to speak regularly to family and friends, you can always use ARC as another source of support.

PREPARING FOR BIRTH

You might have different feelings about giving birth now than you did before the diagnosis. It may be something you are simply 'anxious to get over with' as what comes after is uncertain. Among your concerns may be: the time and place of delivery and how and when your baby will be born. Depending on your baby's condition, you may also be nervous about what he or she will look like. If you feel able to, ask your health care professionals if they can show you some pictures of babies with the same condition to help prepare yourselves. Overall, try not to forget that although your baby may need special care, your wishes as a mother in relation to the birth are also important.

A birth plan

It may be that you will be able to have exactly the kind of birth you had imagined. Whatever your baby's diagnosis, his or her birth can still be special. Depending on your baby's condition, you may have a number of choices about the kind of birth you would prefer. If you had intended to have a home birth, ask if this is still possible. If you have not thought about the arrangements, you might want to think about preparing a birth plan. However, do talk this over with your consultant as in some cases there may be particular considerations that require you to have a Caesarean section.

There are several areas you can cover in your birth plan. You might want to decide on who will be present at the birth and the kind of pain relief you would prefer. You can ask that medical intervention be the minimum necessary, if you wish. If your baby needs to be treated very soon after birth, you can state that you would like to spend time with your baby if possible before he or she has to go for treatment. You can then write your plan down and give copies to your midwife and consultant. They will then know exactly what you want. Bear in mind that producing a plan does not bind you to following it; you can change your mind at any time.

Where will the birth take place?

You will definitely need to know where you are going to give birth. It may be that you can choose between your local and specialist hospital. If you or your baby will require particularly careful monitoring during the birth or special care straight afterwards, you will probably be advised to deliver in a specialist hospital.

If your baby needs to spend time in a special care baby unit, you will be shown round the unit before your due date, so you will know where your baby will be and meet the people who will care for him or her. It may be reassuring to check what will happen were you to go into labour unexpectedly, so you know exactly where you need to go.

If your baby has to stay in hospital

Your baby may need to stay in hospital for treatment straight after birth; depending on the condition, this could be for a short period or could extend to several months. You will need to plan for this, particularly if the hospital is some distance from your home. If you face an extended stay in hospital, make sure you have all the personal items you need and want with you. Have plenty of your own nightwear and toiletries with you as the hospital provision may be limited.

The staff at a special care baby unit (often abbreviated to SCBU) encourage parents to be with their baby as much as possible. There are often ways in which you can be actively involved with your baby's care, so you do not have to feel that this has been taken out of your hands.

You will probably be keen to spend as much time as you can with your baby in the SCBU, but be careful not to neglect your own needs. If you do have other children, try to make sure their lives are not disrupted too much and if childcare is required it is always better, if possible, to call upon someone your children know and trust. There may be someone like this who will be happy to help out, either by taking care of your children in your home or letting them stay with them.

Taking your baby home

It is obviously very important that you are confident about how to care for your baby before you take him or her home with you. Before leaving hospital, you will have an appointment with a consultant or specialist nurse when you can discuss all aspects of looking after your baby. It is useful to draw up a care plan together. As well as everyday care, you may want to ask for any warning signs to watch for and to confirm when it would be necessary to bring the baby in to hospital.

Feeding

You will need to feel confident about feeding your baby before you leave hospital. Your health professionals will help and show you how to use special equipment if it is necessary.

You may want to breastfeed your baby and in many cases this will be problem free. Even if your baby does have difficulty getting milk from the breast, you can express milk that can then be given to the baby.

Talk this over with your midwife or you can contact the National Childbirth Trust if you need further advice on breastfeeding on: **0300 330 0700**.

Your baby may have a condition that means you will need specific advice on feeding. Your consultant paediatrician will be able to help and can put you in touch with the appropriate support organisations if necessary. Make sure you get all the help and guidance necessary - however many times you might need to be shown - to allow you to feel secure about feeding your baby yourself.

Arranging visits from health care professionals

Just because you have left hospital does not mean that you are not entitled to further care and support. There may be a community paediatric nurse in the area who can offer help. Your midwife can come and visit you at home in the first two weeks after birth. If you have had to stay in hospital during this time a community health visitor will come round to see you. The health visitor can make regular visits if that is what you want.

Your GP should be fully informed about your situation. However, communications do sometimes break down, so it may be worth contacting your local surgery to check that your doctor has been kept up to date.

Your own postnatal care

There can be a danger that you overlook your own physical needs in your desire to look after your new baby, particularly if your baby needs special care. After having a baby it is essential that you allow your body to recover and get rest. Looking after a baby with extra needs can be very tiring.

Don't feel that you always have to get other jobs done when your baby is sleeping, use it as time to have a rest yourself. Make sure you eat properly and try to fit in some postnatal exercises too, to help your body back into shape.

If you have any worries about your physical or mental health, talk to one of your health care professionals or contact your GP. You should have a postnatal appointment with your GP or at your local hospital about six weeks after giving birth to make sure all is well with you. Your GP will check that you have recovered physically from the birth and are coping emotionally.

IF YOUR BABY'S FUTURE IS UNCERTAIN OR YOUR BABY IS GOING TO DIE

You may have been told that your baby's chances of survival are slight or that your baby will die. There may be uncertainty about what will happen at or after birth. Because of this uncertainty the pregnancy can become a very precious time as you forge a relationship with your baby. If this is your situation there are certain preparations you may wish to make. It may be upsetting to imagine the worst, but some parents say that preparing for their baby's death gave them something to focus on and allowed them a certain amount of control and choice in what was going to happen.

Some parents like to consider the kind of funeral arrangements they would prefer well in advance and think through ways of remembering their baby. There will be many choices open to you, either through the hospital or privately. Hard as it may be, thinking through what type of burial or cremation service feels right for you and making the necessary arrangements means you can be fully involved in decision-making at a time when it sometimes seems that things are outside your control.

You may also want to consider whether you want to donate your baby's organs, which may be of help to another baby; or tissue samples, which would be used for research. Some parents find comfort in the thought that there may be some benefit to others from the loss of their baby, but it is very much a personal choice and may not feel right for everybody. This is something you can discuss at length with your consultant.

Your hospital will be able to offer various 'keepsakes' ranging from pictures, wrist and ankle bands to swaddling blankets and foot and hand prints. Don't be afraid to ask for whatever mementoes you want. Whatever you choose to keep can then go towards making up a 'memory box' for your baby, to which you can add cards given to you by family and friends. As well as often helping in your grieving process, your memory box is something that you might want to use with other children to help them understand what has happened.

There will be other considerations if you know your baby is going to die after birth. You can state in your birth plan if you would prefer your medical team not to intervene unnecessarily. You may not want a medical team to prolong the life of your baby for too long if he or she is certain to die. You might prefer to care for your baby yourself with the support of your team. You should be allowed all the time you would like with your baby and can ask that other members of your family or friends visit you shortly after the birth. You can also take your baby home if you want to.

You can talk to your team about palliative care (your baby is kept comfortable until he or she dies). Sometimes this can be offered in a children's hospice. The charity Together for Short Lives <https://www.togetherforshortlives.org.uk/> can help with advice and support.

After your baby has died

You may be asked to consent to a post mortem on your baby. This is entirely your choice, but before you decide, talk over exactly what the procedure will involve and check whether information gained could be significant for future pregnancies. If you do agree to a post mortem it can take six weeks or more to get all the results through. Ask for an appointment with your obstetrician soon afterwards to discuss the report. Depending on the findings you may be offered an appointment with a genetic counsellor to talk about the implications for future pregnancies.

Partners

This can be a very testing time for relationships. As a couple, you will both be experiencing strong emotions and may have different ways of expressing them. You may feel the impact of what has happened at different times. Some parents speak of feeling very low at a time when they thought they had come to terms with the situation.

Traditionally, men have not always been encouraged to be as open as women about their feelings. In general, women find it easier to seek support from family members or close friends, whereas men often find it difficult to talk about very painful emotions. The conventional role of father is seen as one of a provider for and protector of the family. He is expected to remain strong and supportive and this could mean that he does not always feel able to talk about his feelings. Fathers may also feel very guilty or angry that they have no control over what is happening to their baby and do not have the power to take the pain away from the mother.

Fathers or partners are often expected to or decide to take on a very practical role. While it may be true that a partner has to deal with day-to-day concerns, this does not mean that the feelings are not present. As most attention is focused on the mother in this situation, it is possible for a father or partner to feel 'left out' or perhaps not consider their own needs. There can also be difficulty in providing emotional support for one another if you are both struggling. This is a time when support from friends or outside family members can be invaluable. Do not be afraid to ask for help; in no way is it a weakness to do so. Often those closest to you will want to do everything they can, but will be unsure what to do for the best and so will be relieved when you ask for help.

Whatever your situation, it can be helpful to keep talking and try to respect one another's way of coping with this stressful time. Sharing an intense emotional experience brings some couples closer, but it can also put a tremendous strain on relationships. If things between you do become particularly difficult, a counsellor or therapist may help. Do contact ARC to discuss this.

Children

If you have other children, they will be aware that you are stressed and will be imagining all sorts of reasons for this, which could be frightening for them. For your benefit as well as theirs, you will need to decide when and what to tell them. Much of what you say will depend on their age and the nature of the condition, but do try to be as honest as you possibly can, using language they will understand. Be ready to be faced with a range of emotions which could vary from anger that their new brother or sister is not going to be the ideal playmate through to anxiety and guilt that perhaps they caused this to happen because they did not want a new family member. This will not be easy for you when you are feeling vulnerable, but try to be as reassuring as you can.

Although your children may be affected by your distress, it can help if they are kept as near as possible to their usual routine, as this will help them feel secure. This may take some organising if their mother faces a prolonged stay in hospital or a number of visits to a specialist hospital some distance from home. If you are going to be away from home for long periods, it may help to ask someone your children like and trust to look after them. Perhaps a friend or family member could help or even stay with the children in your home so as not to cause too much disruption. It can also be useful for children to have someone else to talk to as you may have difficulty dealing with some of their reactions when you are in need of support yourself. You may also want to think about alerting their school to what is going on at home, so their teachers can keep a sympathetic eye on them and will be sensitive towards any changes in their behaviour.

Older children may find it easier to understand what is happening, but may not want to speak openly to you about their feelings. This could be through a fear of burdening you further. They may have feelings that they are uncomfortable expressing to you such as a fear or embarrassment at the prospect of having a brother or sister who is 'different'. Make it clear that they can talk to you if they want to but be aware that they might appreciate being able to talk things through with someone outside the immediate family. The charity for siblings of children with disabilities, SIBS, may be helpful: <https://www.sibs.org.uk/>

Other family and friends

You may want to think about what you are going to tell those outside your immediate family. It will depend on your individual circumstances, but some people decide to send an email or text to those they want to know with brief details of what is going on. If you work, you might want to ask someone you trust to tell others at your workplace, to save you the stress of doing so. It will be important that your employers and perhaps some colleagues know something about what is happening, so they will be aware of the fact that you may need extra time off work, sometimes be upset or have difficulty concentrating.

Other people

It is quite common for people you do not know well or strangers to comment or question you when they notice you are pregnant. This could happen in all sorts of places, at work, at the school gates or at the supermarket. It is almost always well meant, but may take you by surprise. It is useful to think about how you might respond to such enquiries in advance, so that you are prepared.

GETTING HELP AND SUPPORT

Remember you do not have to be alone to deal with all we have covered in this booklet. There may be a specialist organisation, for parents of children with the condition your baby has, which can offer you ongoing advice and support. Ask Contact or see if your hospital can help refer you to someone.

You can also contact ARC at any time during your pregnancy. We are not qualified to give medical advice, but can help you obtain the advice or information you need. We are also here to listen; it can sometimes be helpful to talk through your feelings during the pregnancy with someone who is outside your immediate circle. On the ARC helpline you will be able to speak to someone who has time for you but will not be falsely reassuring. Please feel that you can use ARC as an extra source of support whenever you want to.

GLOSSARY OF TERMS

What follows is not a comprehensive medical glossary, just some terms you may hear used by health professionals or see written down.

A(n) used at the beginning of many words to mean that something is missing, lacking or it can simply mean *not*, e.g. **acardia** (absence of the heart), **amorphic** (lacking definite shape) **atypical** (not usual).

AC (abdominal circumference) measurement used in ultrasound to help calculate a baby's gestation.

agenesis (a-genesis) in medical terms, agenesis means absence of something. **Agenesis of the corpus callosum** In this case, a part of the brain that joins the two hemispheres together is missing. **Renal agenesis** means the kidneys are missing.

anomaly a malformation or abnormality in any part of the body.

aplasia total or partial failure of development of an organ or tissue.

atresia (atresia) absence or abnormal narrowing of a body opening, e.g. **duodenal atresia**, the narrowing of part of the small intestine that consequently causes an obstruction.

autosome any chromosome that is not a sex chromosome.

autosomal dominant describes a situation in which the person possessing an altered gene will show characteristics of the genetic disorder and has a 50% chance of passing it on to a child.

autosomal recessive describes a situation in which the carrier of an altered gene does not show any characteristics of the disorder. The baby of two carriers of the altered gene has a 25% chance of inheriting the disorder.

BPD (biparietal diameter) measurement of two bones in the skull used by ultrasonographers to help calculate a baby's gestation.

cardiology the science concerned with the structure, function and diseases of the heart. *cardiologist*.

centile chart chart used to assess a baby's/child's physical development. The lines of growth on the chart are called centiles. The number of the centile (from one to a hundred) predicts the percentage of babies/children who are below that measurement for a particular age. E.g. if a baby falls within the 10th centile, it means 10% of all babies will be smaller and 90% will be bigger. A baby would be expected to stay within the same centile as it develops.

cephalic (kefallic) relating to the head.

chromosome a rod like structure present in all the body's cells (apart from red blood cells) which stores genetic information. Normally, humans have 23 pairs of chromosomes. The sperm and ova contain 23 each which combine at fertilization to create 46. Each chromosome pair is given a number to describe it, from 1 to 23 (number 23 is the pair of sex chromosomes, XX or XY). e.g. Down syndrome describes a condition in which the baby has three of chromosome 21 rather than the usual pair, so it is also known as Trisomy 21.

congenital (kongenital) describing a condition present from birth, not necessarily inherited.

consultant a highly trained specialist in a branch of medicine who accepts total responsibility for patient care.

CRL (crown rump length) measurement of baby from top of the baby's head to its bottom used in ultrasound to help calculate gestation.

cyst an abnormal sac or closed hole containing liquid or semi-solid matter. *cystic*.

cystic hygroma cyst on lymphatic vessels, usually around the head and neck region. These cysts can vary enormously in size, from a small growth on the neck to a very large mass of growths which can seriously inhibit eating.

deletion genetic material is missing on a chromosome.

diaphragmatic hernia (dyafrematic) sometimes referred to as a **congenital diaphragmatic hernia (CDH)** a hole in the diaphragm which the bowel can pass through. This could affect the development of a baby's lungs if the bowel goes up into the chest and then squashes against the heart and lungs.

dys- used at the beginning of many words to mean difficult, abnormal or impaired, e.g. dysplasia.

dysplasia (displayia) abnormal development of skin, bone or other tissues.

fetoscopy a special fibre optic instrument is passed through the abdomen of a pregnant woman to examine the baby. If required, it is usually done in the 18th-20th week of pregnancy.

gene the unit of a chromosome through which particular characteristics are inherited from one or both parents.

FL (femur length) measurement of thigh bone used in ultrasound to help calculate a baby's gestation.

genetic counsellor a health professional who specialises in advising parents about inherited disorders and the likelihood of becoming affected or having affected children. He or she can also give information on what may be available in order to prevent, diagnose and manage such conditions.

gynaecology the study of diseases of women and girls, particularly those affecting the female reproductive system. *gynaecologist*.

haem- used at the beginning of words to mean blood, e.g. *haemorrhage* (bleeding) *haemophilia* (inherited disorder in which blood clots very slowly).

hereditary describes how characteristics are transmitted through families within the chromosomes of the fertilising egg and sperm.

hydro- This is used at the beginning of many words to mean water or watery fluid, e.g. *hydrocephalus*.

hydrocephalus (hydrokefalus) an abnormal increase in the watery liquid, known as cerebro-spinal fluid, in the ventricles of the brain. In babies this will make the head enlarge. Sometimes it is referred to as 'water on the brain'.

hydrops an abnormal build-up of fluid in body tissues or cavities **hydrops fetalis** describes severe swelling that develops before birth most often because of anaemia.

hyp(o) - This is used at the beginning of many words to mean deficiency, lack or small size, e.g. *hypoplasia*. When speaking anatomically it can also mean under, e.g. *hypodermic* needle - syringe used to inject a substance under the skin.

hypoplasia (hypoplasia) underdevelopment of an organ or tissue.

intra- used at the beginning of words to mean inside or within, e.g. *intrauterine* - within the uterus.

karyotype this is done after an invasive test. In the laboratory all the baby's chromosomes are carefully analysed under a microscope and organised in numbered order. They will be able to detect chromosomal disorders by doing this.

IUGR (intrauterine growth retardation) a condition which leads to a baby being born extremely small for its gestational dates.

magnetic resonance imaging (MRI) a way of analysing body tissues by using radio waves and a magnetic field. It is particularly useful for examining the central nervous system and musculoskeletal system. It is a non-invasive diagnostic tool and it does not use potentially harmful radiation as in x-rays.

Micro-array test done on a sample from CVS or amniocentesis which can be used to find tiny bits of extra or missing genetic information (known as copy number variants or CNVs) that are too small to be detected by traditional karyotyping.

microcephaly (microkefali) a condition in which the brain is not fully developed and the head is very small in relation to the rest of the body.

monosomy the absence of one of a pair of chromosomes.

mosaicism where a genetic or chromosomal abnormality occurs in some body cells. The proportion of normal to abnormal cells will determine how serious the condition is.

neonate used to refer to a baby in the first four weeks of life. *Neonatal, neonatologist*.

neural tube the structure in an embryo from which the brain and spinal cord form. **neural tube defects** (NTDs) occur when this does not form properly. There are several types of NTD, including spina bifida.

neurology the branch of science that deals with the structure, functioning and diseases of the nervous system. *neurologist*.

obstetrics the branch of medical science concerned with pregnancy, childbirth and the period of about six weeks following the birth, when the reproductive organs are recovering. *Obstetrician*.

olig- (oligo) used at the beginning of words to mean few or not enough, e.g. *oligohydramnios* (see next entry).

oligohydramnios (oligohydramnios) a condition in which the amount of amniotic fluid is unusually small. It often means the baby's growth is affected.

omphal- (omphalo-) concerning the navel or umbilical cord.

ost- (oste-,osteo-) used at the beginning of words to refer to bone, e.g. *osteoporosis* - loss of bony tissue, causing brittle bones.

paediatrics the branch of medical science concerning child health. *Paediatrician*.

perinatal relating to the period that starts a few weeks before birth, includes the birth and a few weeks afterwards.

-plasia used at the end of words to mean formation or development, e.g. *dysplasia*.

poly- used at the beginning of many words to mean many; multiple or excessive, e.g. *polycystic* describes multiple cysts.

polyhydramnios (polyhydramnios) a condition in which there is too much amniotic fluid. It can lead to premature delivery and can also indicate other anomalies.

postpartum concerning the few days after birth.

pulmonary (pullmoneri) to do with the lungs.

renal (reenal) to do with the kidneys.

shunt a surgically inserted tube that can divert blood or other fluid from one of the baby's bodily channels to another. A shunt can be inserted in some cases of hydrocephalus to drain the build-up of cerebro-spinal fluid.

stenosis the abnormal narrowing of a passage or opening, such as a blood vessel or heart valve.

syndrome combination of symptoms and signs grouped together to form a disorder.

translocation happens when there is a rearrangement in which a piece of one chromosome is transferred to another with a different number.

triploidy the presence of a full extra set of chromosomes.

trisomy (trysomi) the addition of a complete extra chromosome to a pair. Down syndrome is also known as Trisomy 21, as there is one extra chromosome 21.

ventricle (heart) either of the two lower chambers of the heart which both have thick muscular walls. The left ventricle is thicker than the right and pumps blood into the aorta; the right ventricle pumps blood into the pulmonary artery.

ventricle (brain) one of the four cavities which are filled with fluid in the brain.

ventriculomegaly the ventricles of the brain are enlarged.

If this information has raised any questions or concerns, please contact a member of the ARC helpline team on 020 7713 7486 or info@arc-uk.org

OTHER USEFUL CONTACTS:

The number of condition specific support organisations in existence is too extensive for all to be listed here. **Contact** and **Genetic Alliance UK** will often be able to signpost you to relevant sources of support.

British Association for Counselling and Psychotherapy: www.bacp.co.uk

Contact: for families with disabled children www.contact.org.uk

Genetic Alliance UK: umbrella organisation for over 200 charities supporting families with genetic conditions www.geneticalliance.org.uk

NCT: general support for new parents www.nct.org.uk

Sands: support around stillbirth and neonatal death www.sands.org.uk

Sibs: for siblings of disabled children and adults www.sibs.org.uk

Together for Short Lives: for children living with life threatening or life limiting conditions
www.togetherforshortlives.org.uk

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