Early Support

for children, young people and families

Information for if your child has a rare condition

About this resource

In this resource you will find information on:

- what a rare condition is
- getting a diagnosis for your child
- getting information on your child's rare condition
- working with practitioners involved in your child's care
- who can help
- tips and strategies
- where you can get further information and support

This resource is for parent carers with children who have been diagnosed with a rare condition or who it is suspected have a rare condition.

This resource was developed by Contact a Family for Early Support.

Early Support

Early Support is a way of working, underpinned by 10 principles that aim to improve the delivery of services for disabled children, young people and their families. It enables services to coordinate their activity better and provide families with a single point of contact and continuity through key working.

Early Support is a core partner supporting the implementation of the strategy detailed in Support and aspiration: A new approach to special educational needs and disability, the Government's 2011 Green Paper. This identified Early Support as a key approach to meeting the needs of disabled children, young people and their families.

Early Support helps local areas implement the Government's strategy to bring together the services families need into a single assessment and planning process covering education, health and care. Early Support provides a wide range of resources and training to support children, young people, families and service deliverers.

To find out more about Early Support, please visit www.ncb.org.uk/earlysupport.

Where a word or phrase appears in colour, like this, it means you can: look them up in the Glossary at the back of the resource; find contact details for the organisation or agency highlighted in the Further information and useful links section; or find out more in the Who can help section.

Explanation of the term parent carer

Throughout this resource the term ‘parent carer’ is used. It means any person with parental responsibility for a child or young person with special educational needs or disability. It is intended as an inclusive term that can cover foster carers, adoptive parents and other family members.

Contents

What is a rare condition?

The European Union defines a condition as rare if it affects fewer than five people in every 10,000. EURODIS, a support organisation for people living with a rare condition in Europe, estimates that there are between 6,000 and 8,000 known rare conditions, and this number is increasing as new rare conditions are identified.

Rare conditions can affect both children and adults, and can occur at any time of life. Most rare conditions are life-long and some are progressive (may get worse over time). In some cases, rare conditions can be life-threatening.

How rare is rare?

Although each rare condition affects relatively few people, the total number of people affected by a rare condition is quite large. Rare Disease UK, a national alliance for people with rare diseases, estimates that there are 3.5 million people living with a rare condition in the UK. Most rare conditions (around 75 per cent) affect children.

If your child has a rare condition you may feel quite alone. You may be told that your child is the only one in the country to be affected by the condition. While this is occasionally true, it may be that the person who told you this has not seen a child with the condition before, or even heard of it.

However, there may be other families in the UK, or in other countries, with a child who is affected by the same condition. Nowadays, with the internet and support groups making international contacts, it is more likely that you can get in contact with others who understand what you are going through.

"Nobody knows what you're going through, or describes it better than another parent in a similar situation." Parent

Getting a diagnosis

It can take some families a long time to get a diagnosis for their child's rare condition. This can be frustrating but there are several reasons why it is hard to get a quick diagnosis.

Symptoms and presenting features of some conditions can be similar for different conditions

Some rare conditions have symptoms such as muscle weakness, tiredness, pain, vision problems, dizziness or coughing. These are known as non-specific symptoms because they can appear in many different conditions. Practitioners are trained to look at more common causes of symptoms first. Once common conditions are ruled out, they may consider rare conditions as the cause.

Identifying the rare condition is difficult

It may seem obvious, but the fact that a condition is rare makes it very difficult to recognise. You may see several specialists before you finally find one who knows something about your child's symptoms and condition. Some conditions are so rare that only a clinical geneticist will be able to identify them after genetic testing is carried out.

Rare condition symptoms are unusual

You would think having an unusual symptom would make it easier to get a diagnosis. This may be the case if the specialist examining your child is familiar with the condition that causes the symptom. If they don't know about the condition that causes the symptom, then it is likely your child will be referred to another specialist who may know something about the condition.

Referral to specialists takes time

It is difficult to get an appointment with some specialists who deal with rare conditions. Sometimes you may have to wait three to six months for an appointment. It can be frustrating to wait a long time only to be told by the specialist that they are not sure what condition your child has and that you need to be referred to another specialist. Keep trying until you get to see somebody who can help.

Your child's symptoms may not be typical

You child may have a rare condition, but their symptoms do not fit the classic or typical picture of the condition. They may have symptoms that don't usually signify the condition, or may not have all the symptoms that are expected with the condition. Health practitioners may hesitate to diagnose your child with a rare condition because of this.

If you don't feel that all avenues to get a diagnosis for your child have been explored fully, be persistent. Go back to your GP to ask for a second opinion. Some families do not get a diagnosis for their child for some time and others may never get a diagnosis – see the Early Support resource Living without a diagnosis at www.ncb.org.uk/early-support/resources.

Even if you do not have a diagnosis, you and your child are entitled to help and support. For more information, please call Contact a Family's freephone helpline.

"Try to enjoy your child. It is easy to focus on all their problems and forget to enjoy watching them grow up." Parent

Getting information on your child's condition

Contact a Family

If your child's condition is not well known, it can be difficult to get information about it that is easy to understand. Contact a Family produces an online directory. This contains descriptions of hundreds of different medical conditions, including rare disorders. The information is designed to be accessible to a wide range of people. Each entry is reviewed by a specialist on the condition and details of 400 UK support groups are listed.

Even if your child's condition is not in the Contact a Family directory, there may be information in their internal records. Call Contact a Family's freephone helpline to speak to an adviser about finding information.

It is important to remember that children with the same condition can be affected in different ways. You may see a description of a condition that is the worst-case scenario. It is a good idea to take any information about the condition that you find along to your next appointment with your specialist to discuss how it applies to your child.

Other sources of information

Support groups
Condition support groups will sometimes produce information on the condition they support. Groups normally work with a medical practitioner to make sure their information is accurate, but it is always worth checking with the group that their information is approved and who approved it.

Orphanet
Orphanet is the largest rare condition website in Europe. It contains information on around 6,000 conditions and is free to use. It also has information about orphan drugs, specialised centres for rare conditions, laboratories that offer diagnostic tests and clinical trials taking place in Europe. In addition, it lists European patient organisations for rare conditions.

The National Organization for Rare Disorders (NORD)
NORD is a federation of voluntary health organisations dedicated to helping people with rare conditions. Its Patient Information Centre has an 'ask the nurse' and 'ask the genetic counsellor' service. It also has a database of rare conditions and patient support organisations. This is an American website so not all the information will apply to the UK.

"*When they said that my daughter had a rare condition that would affect her growth, I was stunned and totally knocked back by the news. The worst thing was not knowing anything about her condition. I started to panic, thinking that she would never go to school, have a boyfriend or get married*." Parent

Working with health services

Local services

These services will include your GP, the paediatrician at your local hospital and other services you receive near your home, such as physiotherapy, speech and language therapy and occupational therapy.

You may find that practitioners in your local health services (physiotherapy, speech and language therapy and occupational therapy) do not know a great deal about your child's rare condition. As they support children with so many different conditions, it is impossible for them to know about every condition. It may be helpful to get a letter from your specialist (this may be your clinical geneticist) to explain how your child is affected by their condition and what support they need.

As a parent carer, you may have become an expert on your child's condition. If you have found reliable information, you may want to take a copy along to appointments and leave it with local health practitioners. Leave a reasonable amount of time for the information to be digested; a busy health practitioner see lots of patients in one day and may need some time to read through what you have left them.

As a parent carer, your depth of knowledge about the rare condition and knowledge of your own child's needs is a real asset. By working in partnership with practitioners and sharing your knowledge you can get the best outcome for your child.

Specialised services

If practitioners suspect that your child has a rare condition, and there is not the expertise in local health services to help, you may be referred to a specialist hospital, for example, Great Ormond Street Hospital in London, or Birmingham Children's Hospital.

Specialist hospitals deal with much rarer conditions, so by visiting one of only a few such hospitals available in the UK, you are more likely to meet a specialist who has seen other children with the same condition. Families tell us they like to see a specialist who knows about their child's condition, but there is often a lot of travelling involved to get to specialist centres.

For some rare conditions, specialist clinics have been set up. This means that people affected by the condition can see all the specialists they need to in one appointment under one roof. Specialist clinics/centres are usually run by the NHS Specialised Services in England.

You can search for rare condition specialist centres in Europe on the Orphanet website. Your paediatrician may also be able to find out if there is a specialist centre for your child's condition. Support groups are often aware of specialist centres and their members may be able to recommend specialists whom they have found to be good to work with.

Some parent carers find that they are able to manage their child's condition effectively by using specialised services when necessary and attending local services for routine assessments, check-ups and tests. If you can do this, you will need to find out what tests can be done locally. You can also encourage the specialist centre your child visits and the local service (such as your paediatrician) to share information about your child.

Access to medicines

Before medicines can be used to treat conditions they must be approved (otherwise known as licenced). To be approved for a certain use, the medicine must be tested in a number of trials to show it is safe and effective. For a medicine that treats a common condition it is easy to run a lot of trials, but for a medicine that treats a rare condition it is hard, as fewer people have the condition. Drugs or medicines developed for rare conditions can be called orphan drugs. For more details on orphan drugs go to the section What is being done to support people with rare conditions?

Using medicines when they haven't been approved for a certain group of people or for a certain condition is called 'off-label' use. This means that the main use of the treatment is for something else, but practitioners think it may benefit another group of patients. With close supervision the medicine may be tested in this group.

Medicines for children are quite often used off-label because trials are carried out with adults. So if your child has a rare condition, the medicines that your child may be prescribed are likely to be off-label.

As a parent carer you may have discovered some information on off-label medicines that could help your child. If you are a member of a support group, you may also find out information from other parent carers about treatments or medicines that worked for their child.

In some cases, you may be taking along information about a medicine to a health practitioner to discuss whether it would benefit your child. It is important to work in partnership with your child's specialists in these situations. Take your information along and ask to discuss it together. The specialist will be able to help to decide if the treatment is right for your child. All children are different, so what is right for one child may not be for another.

"Try not to worry too much about the future. Try to focus instead on dealing with each day at a time." Parent

Working with education practitioners

A number of other practitioners may be involved in supporting your child. These may include social workers and education staff, such as nursery school workers and teaching assistants/teachers.

It is important that these people have a good understanding of your child's needs and how your child should be supported. You may find it helpful to give them copies of any reliable information on the condition that you have found. It may also be helpful to get a letter from your child's paediatrician or specialist to outline your child's needs or any difficulties they may have in an educational setting, and how these could be supported.

A child with additional learning needs as a result of their rare condition is entitled to get additional help. The main point of contact for a parent carer whose child has special educational needs (SEN), is the school, nursery or other early years setting's special educational needs coordinator (SENCO). If your child is struggling, you may want to meet with your educational setting's SENCO to see what support can be put in place.

You can also ask for your child to be assessed for a statement of special educational needs. The statement will outline the support the school will give them. This system is changing from April 2014. For more information call Contact a Family's freephone helpline.

Each local authority also has a Parent Partnership Service (PPS), which offers information, advice and support to parent carers of children and young people with SEN. The National Parent Partnership Network supports and promotes the work of PPS across England and offers support to parents and carers of children and young people with SEN.

"Try not to focus on the milestones your child has not reached. Focus on those that they have." Parent

Communicating your child's needs

If your child has a rare condition, you may struggle to communicate their needs to health or other practitioners who work with your child. This may be because practitioners do not know a lot about the rare condition. Parent carers often say it is frustrating to have to continually repeat their child's life story. Use of resources such as the Early Support Our family or communication/hospital passports can help.

Early Support Our family

The Early Support Our family can help with coordinating support and sharing information. It can be used to:

- share information about your child's situation with practitioners
- keep track of multiple contacts
- discuss what is most important and then make a plan of what should happen next with everyone who is involved in your child's care

The 'Introducing ourselves' section can be used to write down everything you would like someone to know about your child and your family. This will stop you from having to repeat the same information to different people.

Find out more at www.ncb.org.uk/early-support/resources.

Communication passports

A communication passport or hospital passport is a booklet that is used to pass on crucial information about a child or young person with additional support needs. It contains information about their condition, medications, likes and dislikes, and essential information if an emergency happens. The Foundation for People with Learning Disabilities has a good template for a communication passport in its booklet *An ordinary life.* Bristol Children's Hospital produces a good hospital/communication passport to be used when children are admitted for treatment.

Who can help

Contact a Family

This charity can help in the following ways:

- It provides advice, support and information to any parent carer of a child with additional needs or a rare medical condition through its freephone helpline – in more than 170 languages.
- It offers approved medical information on many conditions, including rare conditions. Visit its online directory at www.cafamily.org.uk/medical-information/conditions.
- It can put you in touch with local and condition-specific support groups, and other families through its online family-linking scheme at www.makingcontact.org.
- It offers a one-to-one family linking scheme for rare conditions in cases where there is no UK support group.
- It helps groups of parent carers to get together and form their own support group – many support groups for rare conditions began this way.

Climb

Climb (Children Living with Inherited Metabolic Diseases) is a UK organisation working on behalf of children, young people, adults and families affected by metabolic disease. It provides condition-specific information, advice and support to families, including linking families together. It also funds educational and primary research programmes and investigates treatments and medical services. See the Useful organisations section for full contact details.
www.climb.org.uk

Genetic Alliance UK

Genetic Alliance UK is an umbrella body that represents many individual patient organisations that support people affected by genetic conditions. It runs several projects to improve the lives of people affected by genetic conditions, some of which are very rare. The alliance provides a lot of useful information on its website, including contact details for regional genetics centres, information on types of inheritance and on types of genetic tests and prenatal diagnosis methods. See the Useful organisations section for full contact details.
www.geneticalliance.org.uk

Unique

Unique is a source of information and support to families and individuals affected by any rare chromosome disorder and the practitioners who work with them. Unique is a UK-based charity but welcomes members worldwide. The charity links families with children affected by rare chromosomal conditions to other families with similar diagnoses for mutual support, and provides family-friendly, medically verified, condition information leaflets. It also has leaflets on different types of genetic tests and analyses. See the Useful organisations section for full contact details.
www.rarechromo.org

"Having someone you can really talk to and who is not emotionally involved – such as a counsellor or health visitor – can be a great help, even if they don't have all the answers."
Parent

Meeting other parent carers

National condition support groups

As a parent carer of a child with a rare condition, you may want to meet other families that are going through similar experiences. In the UK, there are many support groups that help families with a child affected by a particular condition. These are known as condition-specific support groups. Condition-specific support groups may:

- provide information on the condition for families and practitioners
- support families and practitioners who work with them
- link families together
- run fun activities for families, including family fun days
- run medical conferences
- fundraise for research
- advertise research

Contact a Family's online directory is available at www.cafamily.org.uk and lists over 400 condition-specific support groups, including their contact details and the support they offer.

Local parent support groups

Many areas in the UK have a local parent carer support group, where families of children with all kinds of additional needs come together for mutual support and contact. It might be helpful to find out if there is one near you. If your child has a rare condition it is unlikely that another child in the group would have the same condition. However, a lot of the issues you face on a day-to-day basis will be familiar to other parent carers and they often have practical advice to share. You can either call the Contact a Family freephone helpline, or contact your local Family Information Service (FIS) to find local group contact details.

Local groups have the advantage of meeting regularly and locally, which can be important in providing a support network if you need one. Another advantage is that other members can pass on information about support and services available in your area, some of which they may have already used.

"*There was a mum who lived nearby whose daughter had Down's syndrome. I got more information and support from her about getting local services for my child than anyone else*." Parent

"*My child was diagnosed with IgA nephropathy – nobody had heard of it, but our children's centre, through Early Years Equality, found other Asian families who had a child with the condition. It opened up our world and eased our worries to know our child was not alone, nor were we*." Parent

Getting support for you

Having a child who has a rare condition can be emotionally draining. It is important that as a parent carer you get as much support as possible. Useful support networks include extended family and friends. Early Support has a resource called Looking after yourself as a parent, which explores ways to de-stress and build effective relationships with others. Find out more information at www.ncb.org.uk/early-support/resources.

You may find it helpful to set up a circle of support. This is a group of people who meet together on a regular basis to help someone accomplish their personal goals in life. The circle acts as a community around that person (the 'focus person') who, for one reason or another, is unable to achieve what they want in life on their own and decides to ask others for help. The focus person is in charge. They decide who to invite to be in the circle and how the direction the circle's energy is employed, although a facilitator is normally chosen from the circle to take care of the work required to keep it running.

It may be that your circle of support helps you to find out information on your child's condition, diagnostic tests and local services, so you do not have to do it all yourself. The Inclusive Solutions website has good information about this.

"Raising a child with a disability can be hard work, both physically and emotionally. Don't be afraid to ask for help and take help when it is offered." Parent

What is being done to support people with rare conditions?

UK rare disease plan

In 2009 the EU made a recommendation that all member states, including the UK, form a rare condition plan (or strategy). The UK government has committed to producing a strategy to ensure that people affected by a rare condition in the UK can expect to receive an equal level of care wherever they live. The strategy should be devised by the government by 2013. Rare Disease UK and its members have been key in researching what would need to be covered by the plan to deliver the best care for patients.

Orphan drug development – what is it?

Orphan drugs are developed for the purpose of treating a rare condition. They were introduced by the EU because drug companies are not usually interested in developing a drug for so few patients, as the cost of development outweighs the profits they make. Orphan drug designation comes with financial benefits for the drug companies, as well as other assistance and support from the EU to develop the new medicines.

Getting involved in research

For many rare conditions there is no cure, meaning that the effects of the condition cannot be completely reversed. But there may be treatments and therapies that reduce the symptoms your child experiences. If you have a child with a rare condition, you may have heard about research, trials of new medicines/treatments, or new experimental therapies.

Research and trials

Some families may consider becoming involved in research or clinical trials for treatments for rare conditions. The decision to take part will be individual for each family. Always try to find out as much unbiased information as possible before deciding to take part and make sure you inform your child's specialist about what you have found. NHS Choices offers some information on clinical trials and questions to ask before getting involved. The healthtalkonline website has interviews with parents about how they found out about trials, why they decided to take part, or why they did not take part.

You can find information about ongoing clinical trials on the NHS Choices website, or search on the Orphanet website. Some support groups have information about ongoing research. It is worth asking the relevant condition-specific support group about this.

Experimental treatments

Many families will come across information about experimental treatments in their search for possible treatments for their child. An example of this is gene replacement therapy for a genetic condition, where a changed (or mutated) gene is replaced by a functioning gene.

It is important to discuss any information you come across with your specialist. These types of treatments are still fairly new, so scientists and medical practitioners are gathering information about them. Experimental treatments may not be available on the NHS and even if they are there will usually be very strict guidelines on how they are used.

You may be keen to use an experimental treatment on your child, but practitioners may not agree, perhaps because it is unproven or too expensive to be used. If you are looking for information about these kinds of treatments get impartial advice from a not-for-profit organisation and discuss it with the specialist treating your child. Drug companies offering miracle cures may not always be genuine and the cost of treatment can be very high.

Treatment abroad

In some cases, NHS-funded treatment can be found in countries within the European Economic Area (EEA). There are two ways to access NHS-funded healthcare in other EEA countries:

- **The S2 route (or E112)** – This is a direct arrangement between the NHS and the state healthcare provider in the country of your choice. Prior approval is required.
- **The EU Directive on cross-border healthcare (or Article 56)** – Generally, you will have to pay the costs of treatment abroad and then claim reimbursement from the NHS when you return. However, in some cases it may be possible for the NHS to pay directly.

Visit the NHS Choices website for more details. Treatment not funded by the NHS will have to be funded privately by the family.

You will need to think carefully about whether treatment abroad is right for your child. Try to get as much information as you can from an unbiased source before doing this. Support groups may be in touch with members who have had such treatments and it can be possible to speak to other families about their experience. Not all treatments will be suitable for all children so it is important to discuss your intentions with your child's specialist in the UK. They may be able to offer some advice and may also be involved in any follow-up treatment or care.

Treatments outside of Europe will need to be privately funded by the family. NHS Choices lists points to be considered before embarking on treatments abroad and questions to ask your surgeon or dentist.

Alternative therapies and medicines

A complementary therapy is one that can be used as well as, or instead of, conventional western medicine. An alternative therapy claims to be a complete system, which can be used instead of conventional western medicine. Some complementary and alternative medicine (CAM) is now available on the NHS – an example is acupuncture for pain relief. Therapies will only be available on the NHS if there is enough evidence to show that they are beneficial to a group of patients and that they are cost-effective.

Parent carers of a child with a rare condition may come across a long list of therapies that claim to help with certain symptoms experienced by their child. Try to find information on them from unbiased sources, such a not-for-profit organisations. Not all CAMs will be available on the NHS.

You may be keen to use a CAM on your child but practitioners may not agree, perhaps because they are unproven or too expensive to be used. You can find a directory of CAM practitioners on the NHS Trusts' Association (NHSTA) website.

Condition support groups may be a good source of information on CAMs as members who have used them can help you know what to expect. Remember to discuss the use of a CAM with your child's specialist as the CAM may interfere with other treatment your child is having, particularly medication.

Top tips

Parent carers of children with a rare condition can find the number of practitioners they see and appointments they have to attend overwhelming. These tips from parent carers who have 'been there' may be helpful to you:

- Keep a paper trail. Keep copies of all letters, appointments, and test results in a folder. Keep a note of all the phone calls you make as well.
- Be persistent. If you don't feel all avenues to get a diagnosis for your child have been explored then do go back to your GP. You can ask for a second opinion.
- Use the Early Support Our family to co-ordinate support and share information. The 'Introducing ourselves' section can be used to explain your child's needs and the key people in their life, to save repeating your story. Find out more information here: www.ncb.org.uk/early-support/resources.
- Create a communication passport for your child. This can be a series of flashcards that explain things such as your child's health history, what your child can and cannot do, their medications and any health problems they have. These can be taken to appointments or hospital stays and are a quick and easy way of passing on information to the specialist about your child.
- If you find reliable information about your child's rare condition, take a copy to local services that may not be familiar with the condition, such as your GP, social worker or physiotherapist. Leave people enough time to read it and make an appointment to discuss the information at a later date.
- Work in partnership with health and other practitioners to get the best outcomes for your child. Your expertise on the condition and the needs of your child is likely to be a real asset to this partnership.
- Use a combination of local services and specialised services that suits the needs of your family best – this will reduce stress, time and expense by avoiding long, unnecessary journeys. Make sure that you let practitioners know this is your plan.
- If you feel that you need to see a specialist, or if you think you may have somehow got 'lost in the system', don't be afraid to phone to find out. Ask to speak to the doctor's secretary to find out what is happening with your child's appointment.
- Don't be afraid to ask your specialist questions that are concerning you, however silly or insignificant you think they may seem.
- If possible, try to stick with the same practitioners so that your child gets a good level of care all the time from people who understand their needs.

Further information and useful links

Communication and hospital passports

Bristol Children's Hospital

The hospital has a good template for a hospital communication passport, which can be emailed before a child's appointment. It can also be used in other settings.
www.uhbristol.nhs.uk/patients-and-visitors/your-hospitals/bristol-royal-hospital-for-children/information-and-support/hospital-passport
0117 9230000

Foundation for People with Learning Disabilities

The Foundation works to influence government and local authority policies and services so that they better meet the needs of people with learning disabilities, their families and carers. The Foundation's report *An ordinary life* has a good communication passport template.
www.learningdisabilities.org.uk/publications/an-ordinary-life-booklet

Circles of Support

Inclusive Solutions

A team of psychologists and associates who will work with anyone who wants to bring about the real changes in the system that are necessary to move towards a truly inclusive society. Its website aims to provide accessible, up-to-the-minute national and international information regarding training, books and cutting-edge strategies to support those on the front line of inclusive practice. See their useful information on 'a circle of friends'.
www.inclusive-solutions.com/circlesoffriends.asp
inclusive.solutions@me.com
0115 9567305

Research information

Healthtalkonline

This is the award-winning website set up by the charity DIPEx. More than 2,000 people share their experiences of over 60 health-related conditions and illnesses. Visitors can watch (or read) interview clips of parents who have taken part in clinical research, or share reasons why they decided not to.
www.healthtalkonline.org/medical_research/clinical_trials_parents/People/Stories

NHS Choices

NHS Choices is the online 'front door' to the NHS. It is the country's biggest health website and gives all the information you need to make choices about your health. NHS Choices has a good section on its site about clinical trials, including questions to ask before taking part.
www.nhs.uk/Conditions/Clinical-trials/Pages/Introduction.aspx

Treatment abroad

NHS Choices

This has good information on getting treatment abroad, including funding and questions to ask before going ahead:
www.nhs.uk/NHSEngland/Healthcareabroad/plannedtreatment/Pages/Introduction.aspx

Useful organisations

Contact a Family

This charity provides advice, support and information to families with disabled children in the UK whatever the child's disability or additional need.
www.cafamily.org.uk
info@cafamily.org.uk
Freephone helpline: 0808 8083555 (weekdays)

Climb (Children Living with Inherited Metabolic Diseases)

This UK-based organisation works on behalf of children, young people, adults and families affected by metabolic disease. It provides condition specific information and advice and support to families, including linking families together. It also fund educational and primary research programmes and investigates treatments and medical services.
www.climb.org.uk
info.svcs@climb.org.uk
Freephone helpline: 0800 6523181

Early Support

It is a way of working, underpinned by 10 principles that aim to improve the delivery of services for children and young people with additional needs and their families. It enables services to coordinate their activity better and to provide families with a single point of contact, continuity of care and support through key working. Early Support ensures that service delivery is focussed on the child, young person and family, and that services and practitioners work in partnership with children, young people and their families.
www.ncb.org.uk/earlysupport
earlysupport@ncb.org.uk
020 78436350

Early Years Equality

Provides disability equality and other protected characteristic equality support, guidance, advice, discrimination casework and training to children, families, organisations, providers and policymakers across the UK.
www.eearlyyearsequality.org.uk
enquiries@earlyyearsequality.org.uk
0114 2700214

EURORDIS

This is an alliance of patient organisations and individuals active in the field of rare diseases. Its website has information on rare diseases, orphan drugs, other patient experiences, training resources and news.
www.eurordis.org
eurordis@eurordis.org
+331 56535210

Genetic Alliance UK

This UK alliance is an umbrella body that represents many individual patient organisations. It runs several projects, including Syndromes Without a Name (SWAN) UK, and has lots of information on genetics testing and services, including a list of all UK genetics centres.
www.geneticalliance.org.uk
contactus@geneticalliance.org.uk
020 77043141

National Organization for Rare Disorders (NORD)

NORD is a federation of voluntary health organisations dedicated to helping people with rare conditions. Its Patient Information Centre has an ‘ask the nurse’ and ‘ask the genetic counsellor’ service. It also has a database of rare conditions and patient support organisations.
www.rarediseases.org

The National Parent Partnership Network

Supports and promotes the work of Parent Partnership Services (PPS) across England. Parent Partnership Services offer advice and support to parents and carers of children and young people with special educational needs (SEN)
www.parentpartnership.org.uk

NHS Specialised Services in England

This national organisation is responsible for the commissioning of specialised services that help to improve the lives of children and adults with rare diseases or disorders. Commissioning in the NHS is the process of ensuring that health services meet the needs of the population.
www.specialisedservices.nhs.uk
enquiries@nsct.nhs.uk
020 79323939

NHS Trusts’ Association (NHSTA)

NHSTA is an independent association of NHS Trusts, but is not affiliated with the Department of Health. It has a directory of complementary and alternative practitioners on its website.
www.nhsta.org.uk
020 88357006

Orphanet

This is the largest rare condition website in Europe. As well as information on conditions, it has information about orphan drugs, specialised centres for rare conditions, laboratories that offer diagnostic tests, and clinical trials in Europe. Orphanet also lists European patient organisations for rare conditions.
www.orpha.net

Rare Disease UK (RDUK)

RDUK is the national alliance for people with rare diseases. It was established by the Genetic Alliance UK, the national charity of over 140 patient organisations supporting all those affected by genetic conditions. RDUK is lobbying the government for an effective rare disease strategy for the UK.
www.raredisease.org.uk
info@raredisease.org.uk
020 77043141

Unique

Unique is a source of information and support to families and individuals affected by any rare chromosome disorder and practitioners who work with them. Unique is a UK-based charity but welcomes members worldwide.
www.rarechromo.org
info@rarechromo.org
01883 330766

Glossary

Additional support needs – In this and other Early Support resources, additional support needs is used to refer to any child or young person who has a condition, difficulty, challenge or special educational need, whether diagnosed or not, who is likely to need additional support beyond universal services.

Alternative therapy – Any practice claiming to heal that does not fall within the realm of conventional western medicine. It may be based on historical or cultural traditions, rather than on scientific evidence.

Circle of support – A group of people who meet together on a regular basis to help somebody, known as the 'focus person', accomplish their personal goals in life. The members of the circle of support may include family, friends and other community members. They are involved because they care enough about the focus person to give their time and energy to help them.

Clinical geneticist – Practitioners who are trained in genetics and skilled at diagnosing and providing advice about genetic conditions. Geneticists will be able to explain the results of any tests or examinations that you have and how to manage and treat your condition. They may be able to give an estimation of risk to future pregnancies.

Clinical trials – A clinical trial is a particular type of clinical research that compares one treatment with another. It may involve patients or people with no condition or illness, or both. Clinical trials help determine whether treatments are safe, have any side effects and if new treatments are better than standard treatments.

Communication or hospital passport – A booklet that is used to pass on crucial information about a child or young person with additional support needs. It contains information about a person's condition, medication, likes and dislikes, and essential information for an emergency situation.

Complementary therapy – A therapy or treatment that may be used as well as mainstream techniques to treat or alleviate symptoms of a condition.

The European Union (EU) – An economic and political union of 27 member states, which are located primarily in Europe.

Family Information Service (FIS) – Provides a range of information on all services available to parents, helping to support children with additional needs up to their 25th birthday. FIS also holds up-to-date details of local childcare and early years provision in your area.

Gene – The basic physical and functional unit of inheritance. Genes, which are made up of DNA, act as instructions to make molecules, called proteins, which make up our bodies.

Gene replacement therapy – In gene therapy the patient's own stem cells are removed and a healthy, fully functioning gene is placed into them before they are put back into the patient.

Genetic conditions – Conditions caused by changes (mutations) in genes (our body's blueprint) or chromosomes (the structures that DNA is strung into). Genetic conditions will affect a person from birth. Some genetic conditions are passed down from the parents' genes, but others are caused by new changes to DNA or chromosomes.

GP – This is your family doctor. They will usually be the first person you ask for medical help and advice. They can ask for your child to be seen by another specialist.

Occupational therapy – Pays particular attention to hand and eye coordination, perception and manipulative skills. They can advise and provide suitable aids for feeding, dressing, toileting, bathing and play in younger children, and writing skills in older children.

Off-label – The practice of prescribing medicines for treatment for a condition they are not approved for, in an age group they are not approved for (such as children when the medicine has been tested in adults), in a dose they are not approved for, or a form of administration not usually used.

Orphan drugs – Orphan designation is awarded to drugs/methods that diagnose, protect or treat people for a seriously debilitating or serious and chronic condition affecting fewer than five people per 10,000. Financial incentives are given to drug companies to develop these as it is unlikely that the revenue from developing the medicine would cover the investment in its development.

Paediatrician – A practitioner who specialises in looking after babies, children and young people. A paediatrician can coordinate and liaise with other agencies involved in the management, care and education of the child and family.

Physiotherapy – Helps in the management and development of movement skills. There are a number of ways in which children can be helped. These may include exercises to strengthen weak muscles and games to improve coordination and motor skills.

Special educational needs (SEN) – Children with special educational needs have a considerably greater difficulty in learning than others the same age. SEN includes communication, physical, sensory and emotional difficulties that require additional support to help a child reach their full potential.

Special educational needs co-ordinator (SENCO) – A member of staff at a school or in an early years setting who has responsibility for coordinating SEN provision.

Speech and language therapy – Helps children learn to communicate, either through speech or other methods. This therapy can also be used if there are problems with eating, drinking and swallowing.

Social worker – A practitioner who works to protect vulnerable people, enhance relationships and help families to stay together where possible. Social workers enable people to live fulfilled lives as independently as possible. They are based within the local authority.

Statement of Special Educational Needs – This is a document that records your child's additional needs and the help your school must put in place to meet those needs. You can request an assessment of your child's needs, which may result in a statement which is legally enforceable.

Early Support
for children, young people and families

www.ncb.org.uk/earlysupport

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contact a family
for families with disabled children

Early Years Equality
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