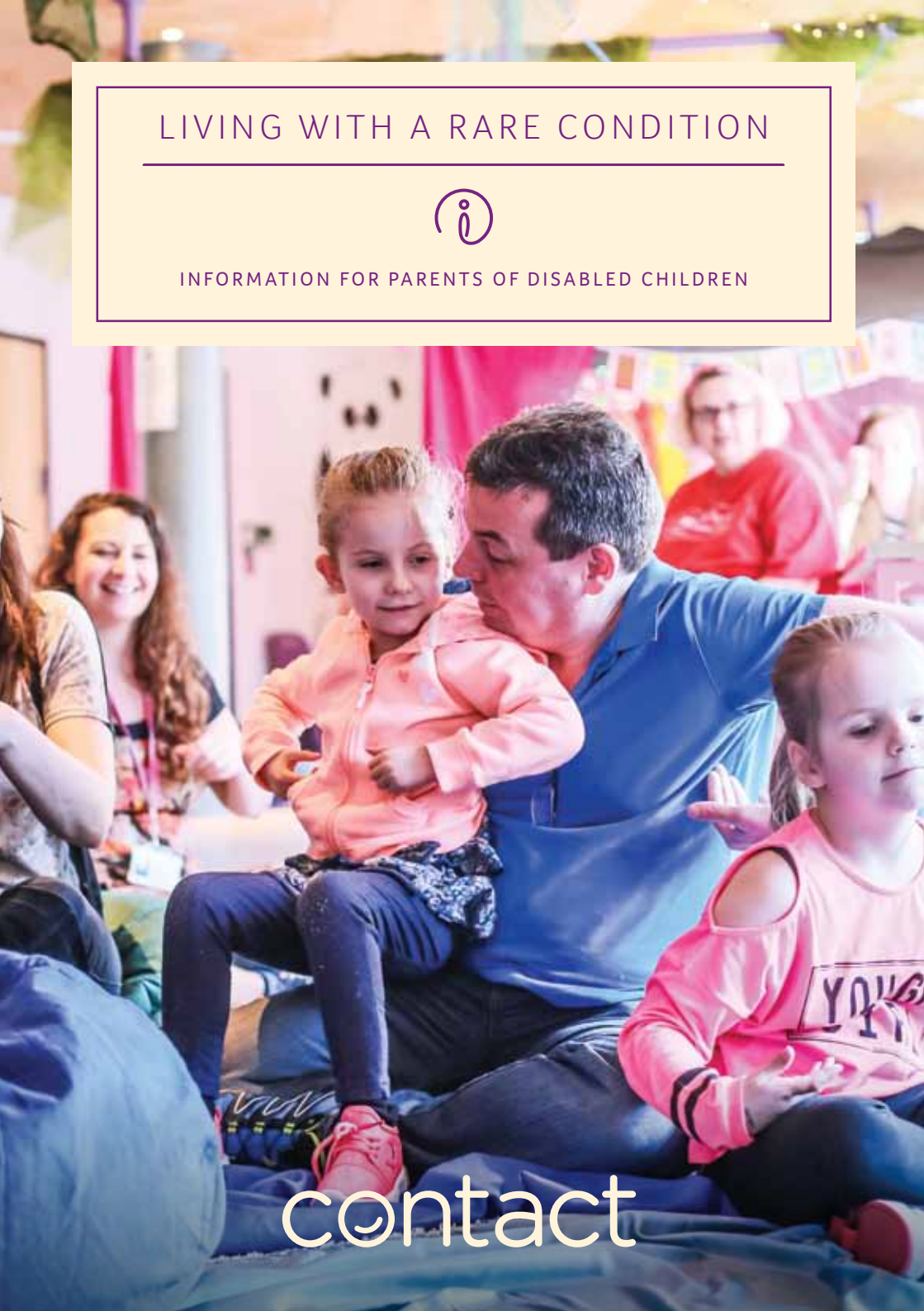
LIVING WITH A RARE CONDITION

INFORMATION FOR PARENTS OF DISABLED CHILDREN



contact

"Realise the potential of your child."

Parent carer

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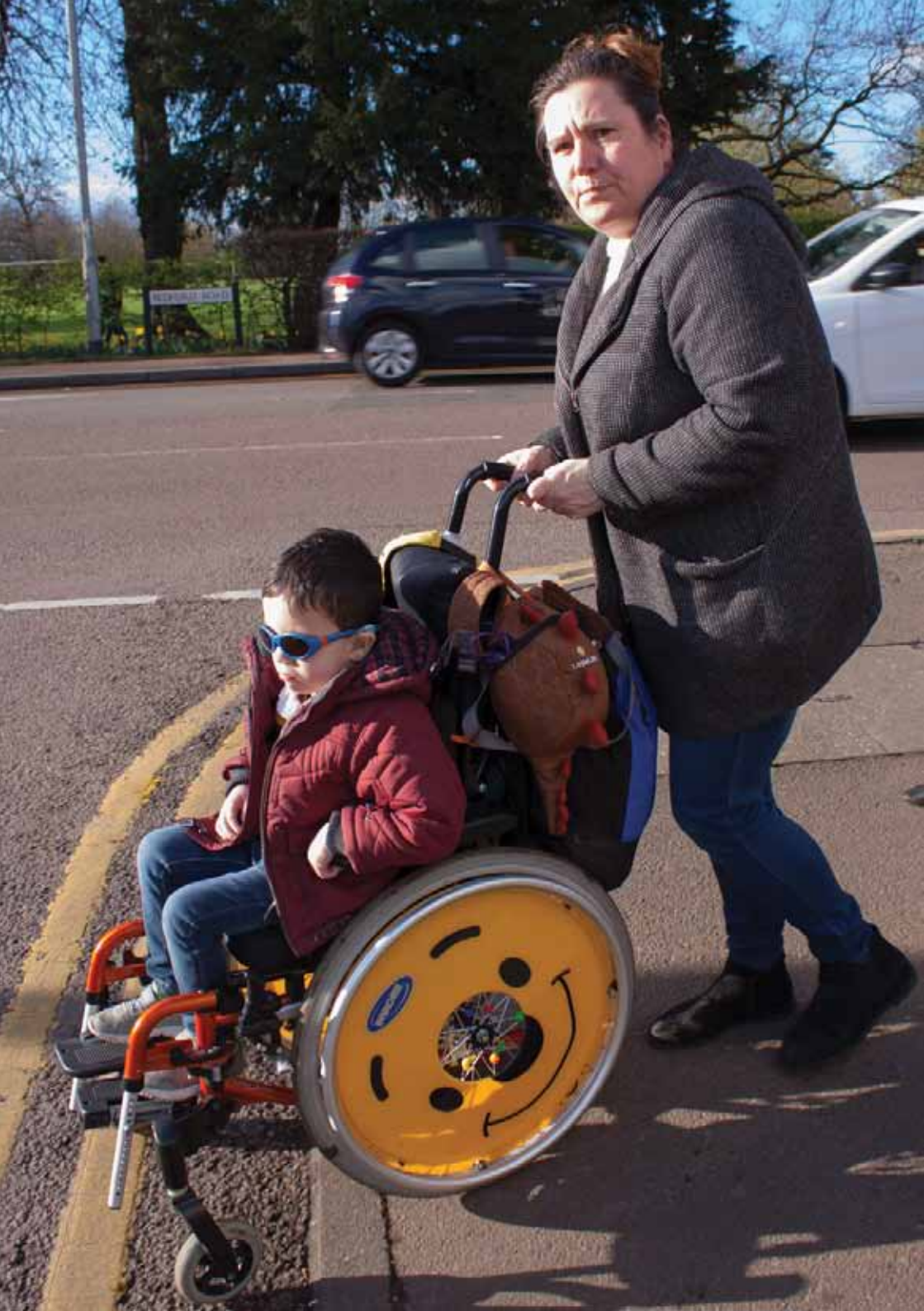


INTRODUCTION

This guide is for parents and carers whose child has been diagnosed with a rare condition, or who think their child may have a rare condition. It contains information on:

- ***what is a rare condition?***
- ***getting a diagnosis***
- ***what help and support you can expect***
- ***meeting other parents***
- ***top tips.***

Around the time of diagnosis, families can go through a range of emotions. Feeling anxious or confused are common reactions, but we can help. We have a number of guides with topics that may be useful to parents who are trying to get a diagnosis for their child. Each guide contains useful information and signposts to further sources of support for parents. They are available free from our helpline 0808 808 3555 or to download from our website:

- ***Developmental delay***
- ***Living without a diagnosis.***

WHAT IS A RARE CONDITION?

The European Union (EU) says a condition is rare if it affects fewer than five people in every 10,000. 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 known as 'progressive' (may get worse over time). In some cases, rare conditions can be life threatening.

EURORDIS, 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 will increase as new rare conditions are identified.

HOW RARE IS RARE?

Although each rare condition affects a small number of people in the population, the total number of people affected by rare conditions is quite large. Rare Disease UK estimates that there are 3.5 million people living with a rare condition in the UK, and 30 million in Europe. Most rare conditions (around 75 per cent) affect children.

If your child has a rare condition, you may feel quite alone. However, there may be other families in the UK, or in other countries, that have a child who is affected by the same condition. With access to the internet and international support groups, it is now possible to get in contact with others who can relate to what you are going through. See page 14.

WHY RARE CONDITIONS CAN BE HARD TO DIAGNOSE

It can take some families a long time to get a diagnosis for their child's rare condition. For some people, this can be frustrating and very upsetting, so it can be helpful for parents to talk to professionals to better understand why an early diagnosis is not always possible. There are several reasons that may delay a quick diagnosis for a rare condition:

- ***Some conditions have similar general symptoms; for example, muscle weakness, tiredness, pain, vision problems, and dizziness or coughing. Doctors are trained to look at common causes of symptoms before they consider rare conditions.***
- ***Some symptoms of rare conditions are unusual – if the specialist examining your child isn't familiar with the condition causing their symptoms, then your child is likely to be referred to another specialist.***
- ***Your child may have a rare condition, but their symptoms do not fit those usually associated with the condition.***
- ***Your child may not have all the symptoms that are commonly seen with the condition.***

> *"My advice for other parents would be; you are entitled to your feelings and do not lose hope, because children with rare conditions can flourish and do amazing things."*
> **Parent carer**

REFERRAL TO SPECIALISTS

This can take time. It can be 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.

IDENTIFYING THE CONDITION

This can be difficult – you may see several specialists before you finally find one who knows something about your child's symptoms and condition.

Our guide **Living Without a Diagnosis** is free from our helpline **0808 808 3555** or to download at **www.contact.org.uk/no-diagnosis**

SOME HELPFUL HINTS FOR APPOINTMENTS

Some parents find it helpful to make a list of all the features their child displays and examples of things they find difficult to take to appointments. You can use a log of your child's development progress to show areas where they may be behind other children.

Parents have told us that it also helps to make a list of questions to ask before they go to appointments. This helps them to remember what they want to ask the specialist. It can also be helpful to have a friend or relative come along to appointments, as they can help make sure you ask all the questions on your list and take notes for you.

WHAT TO DO IF YOU ARE NOT HAPPY

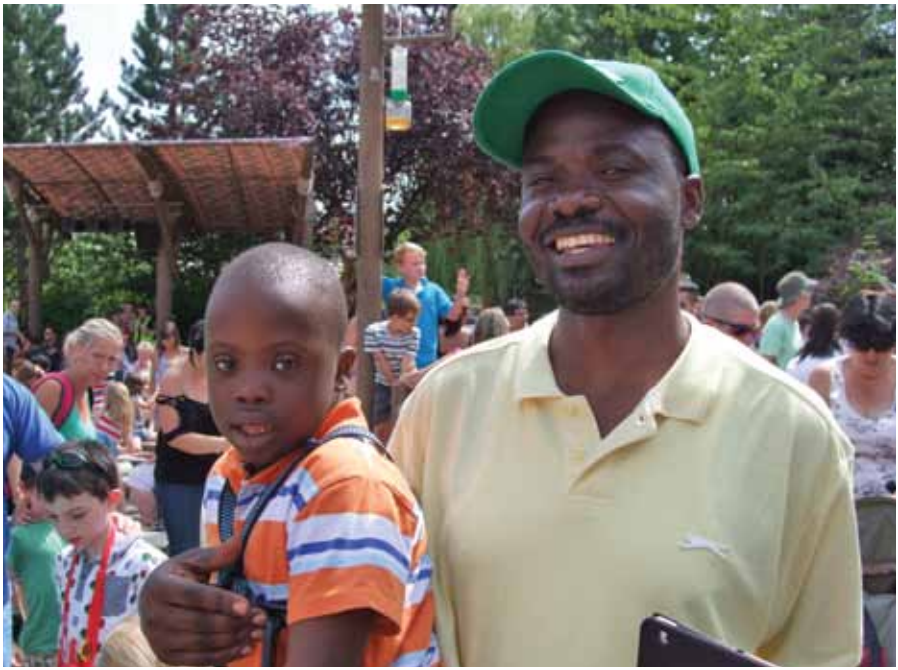
If you feel strongly that all the ways of getting a diagnosis for your child have not been explored, you can ask for a second opinion. You may have a niggling feeling that your child is very similar to other children with a particular condition but other professionals aren't recognising it.

Support groups for the condition may be able to help you find information and the right specialists to make a diagnosis.

If after seeing a consultant doctor or specialist you would like a second opinion, you can go back to your GP and ask them to refer your child to a different specialist. Try to avoid being confrontational but be firm. You can take along the list of things your child finds difficult to help explain the reason why you are asking for the referral. If your GP will not help you get a second opinion, you can ask to see another GP at the same, or a different practice. Contact can also offer support and advice if you are unhappy with your child's care.

"I spent hours looking for information on the internet. In the end I realised I was just torturing myself. My time is better spent with my children."

Parent carer



GETTING INFORMATION ABOUT YOUR CHILD'S CONDITION

If your child's condition is not well known, it can be difficult to get reliable, easy to understand information about it. At Contact, we have information on more than 400 different conditions, including rare conditions, on our website, plus details of UK support groups for them. Support groups for specific conditions sometimes produce medical information for families and professionals. Even if a condition is not listed on our website we may still be able to help, so give us a call.

0808 808 3555 **www.contact.org.uk/medical-information**

MEDICAL INFORMATION ON THE INTERNET

If you're searching for information on the internet, remember that not all online information is accurate and trustworthy so make sure you discuss what you read with a medical professional before acting on it.

Remember that children with the same condition may be mildly affected and some severely affected. Information you find may reflect the most severe cases and not always apply to every child. So take any information that you find to your next appointment with your specialist to discuss how it applies to your child.

Avoid giving out your personal contact details on the internet.

If you're using the internet to find information about medical conditions, you will need to carefully evaluate what you find. Here are some tips to help you judge if a source is trustworthy:

- **Consider who wrote the information.** Charity and government sites are unlikely to be biased, but companies may want you to buy something.
- **Reputable organisations** will provide their address, a way to contact them and an 'About us' section where they describe their purpose.
- **The writing should be free from spelling errors,** grammatically sound and objective and balanced in tone.
- **The page should list the author's name,** their qualification to write on the subject and, in some cases, how the text has been verified.
- **You should be able to find a date on which the information was published or updated;** this helps determine the timeliness of what you're reading. For example, Contact's medical texts carry endorsements in the following form: *Last updated February 2018 by Dr R Gibbons, Professor of Clinical Genetics, Weatherall Institute of Molecular Medicine, John Radcliffe Hospital, Oxford, UK.*

SUPPORT FROM OTHER PARENTS

As a parent of a child with a rare condition, you may want to meet other families that are going through similar experiences. There are many UK support groups that help families with a child affected by a particular condition. Condition support groups may:

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

At Contact we have details of more than 400 condition support groups on our website and hundreds more on our database.

Search on the medical information section of our website at **www.cafamily.org.uk/medical-information** or call our helpline if you can't find one, we may still be able to help.

LOCAL PARENT SUPPORT GROUPS

Many areas in the UK have a local parent support group, where families of children with all kinds of additional needs come together for mutual support and contact.

If your child has a rare condition, it is unlikely that another child in a local support 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 parents and they will have practical advice to share.

Local groups have the advantage of meeting regularly, which can be important in providing a support network if you need one. You can call our free helpline to get information on local support groups.

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

CONNECTING WITH PARENTS ONLINE

For parents whose children are affected by rare conditions, reaching out to others online can be particularly useful, as many will have been told by doctors that there are only a small number of cases in the world.

You may find that someone else's experience helps you to deal with issues you too are experiencing. By talking to others you may find out about treatments, new therapies or research projects that are being conducted to learn more about the condition your child is affected by. See page 21 for more information about this. Always use trusted websites, like Contact's online community, www.contact.org.uk/community, or the ones below.

Rare Connect.org

A safe, moderated platform where rare condition patients, families and patient organizations can develop online communities and conversations. You can also find out about any advancements in understanding a condition, and progress towards effective treatments.

www.rareconnect.org/eng

Rare Revolution

Digital magazine for rare conditions. They also have a closed Facebook group so that people affected by rare conditions can share experiences safely.

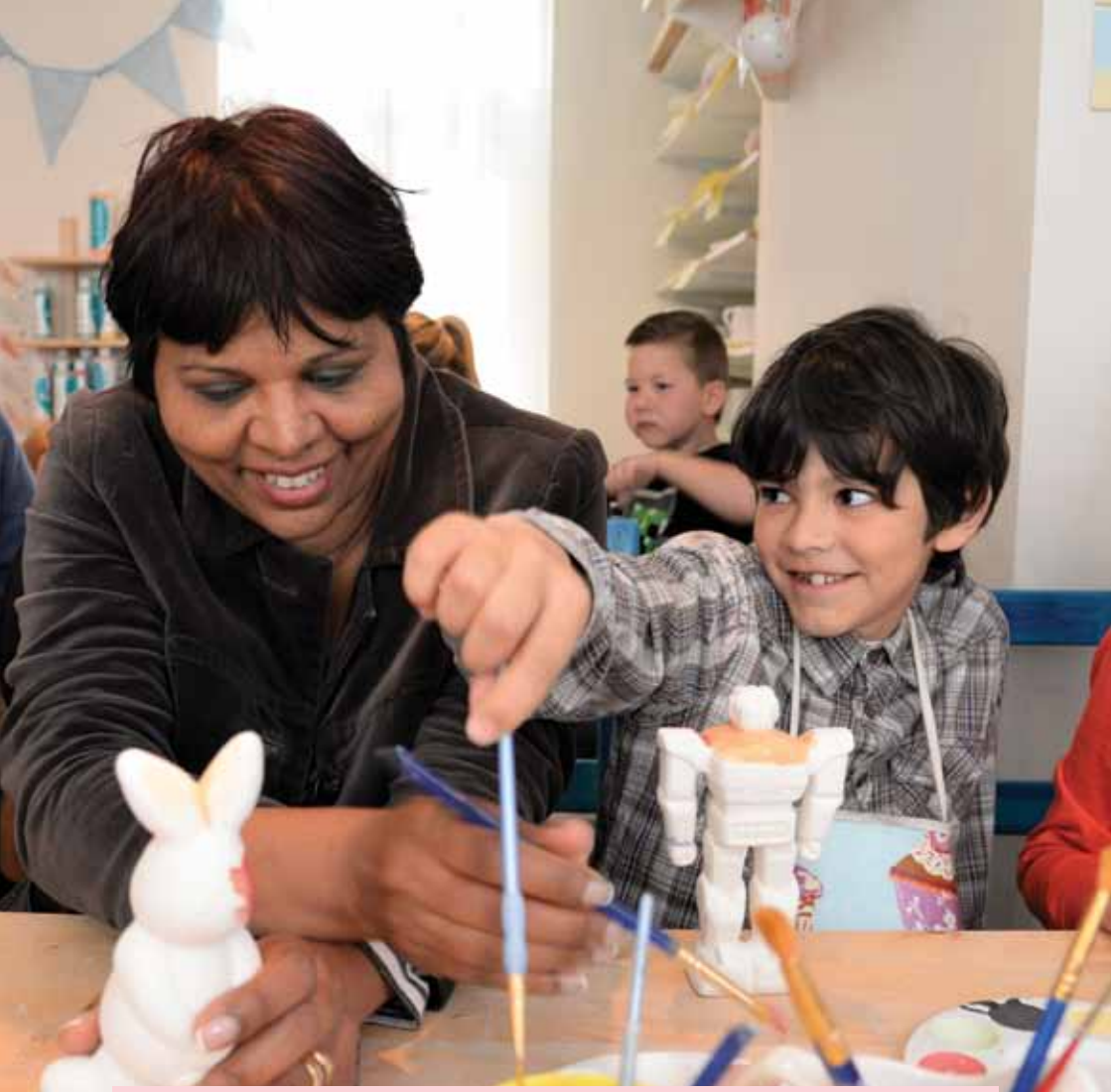
www.rarerevolutionmagazine.com

Facebook may also be a good way to meet other parents – there are lots of online support groups for specific conditions and there is the option of closed or private groups. You can set up your own group if there isn't one, and then other parents can find you. But do remember to check if there is already an established group you can contact – they may already have useful information and advice to share.

TOP TIPS FOR STAYING SAFE IN ONLINE COMMUNITIES

- **Don't over share personal information on a public forum.** For example your email address, telephone number, the school your child attends or specific local services you access. Find a way to communicate that information privately, only to its intended recipient.
- **Use a pseudonym when discussing health information online** and be aware of the data-sharing policies of websites where you share personal information.
- **Protect yourself by customising your privacy options.** You can limit who can see various aspects of your personal information online. Even if you have been a user for some time, log onto your account and view and adjust the privacy settings – new settings are added over time.
- **Consider the source of any information and check with your doctor before making any treatment changes.** Even other well-meaning parents can give out treatment information that could prove harmful.
- **Be careful when sharing your child's full name online.** Something they might not mind at age ten might be something they are not comfortable being available to the public at age eighteen.
- **Be careful sharing photos of your child online too, for the same reason!**

"One of the mums I contacted gave me some useful information about specialist milk for my daughter. I spoke to my dietician who sorted it all out for me. Happy days!"



"The communication passport is amazing. It is simple, short and sweet and gets everything that needs to be said across in a very simple way. I don't just use it at the hospital – I use it everywhere I go, including school. It is so much easier to use this little book than the reams of notes that I used to carry around before."

Parent carer

WORKING WITH HEALTH SERVICES

LOCAL SERVICES

These services will include your GP, a paediatrician at your local hospital and other services your child might receive near your home such as physiotherapy, speech and language therapy and occupational therapy. You may find that professionals in your local health services do not know much about your child's rare condition. It may be helpful to get a letter from your specialist to explain how your child is affected by their condition and what they need support with.

If you have found reliable information on your child's condition, you may want to take a copy along to appointments and leave it with local health professionals. Give professionals a sensible amount of time to read and digest the information. A busy professional may not have time to read a document when you give it to them. You could offer to go back at a later date to discuss the needs of your child.

SPECIALISED SERVICES

If professionals 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. At specialist hospitals you are more likely to meet a specialist who has seen other children with the same condition. An example of a specialist hospital is Great Ormond Street Hospital for Children.

Families tell us they like to see a specialist who knows about their child's condition, but this may involve a lot of travelling 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 place.

Some parents find that they are able to effectively manage their child's follow-up care by using specialist services when necessary, but attending local services for routine assessments, check-ups and tests. It is important to let your specialist centre and local health care centre know about your plan to do this. Information will need to be clearly communicated between both places.

It is important to work in partnership with health and other professionals to get the best support in place for your child. Your expertise on the condition and the needs of your child is important to this partnership.

COMMUNICATION PASSPORTS

Parents often say it is frustrating to continually repeat their child's life story to multiple health workers who may not know about their condition.

A communication or hospital passport is a booklet that you can use to pass on crucial information about a child or young person with additional needs. It contains information about their condition, medications, likes and dislikes and essential information if an emergency happens.

If your child doesn't have a communication passport, then Bristol Children's Hospital has a good template for a hospital passport. Visit its website and search for 'passport': **www.uhbristol.nhs.uk**

Mencap has a downloadable hospital passport for children with a learning disability on it's website: **www.mencap.org.uk/advice-and-support/health/our-health-guides**

ACCESS TO MEDICINES

For medicines to be used to treat conditions they must be approved for use – also known as getting a licence. To get approved for a certain use, the medicine must be tested in a number of trials to show it is safe and effective.

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 something else, but doctors think it may benefit another group of patients. Medicines for children are quite often used off-label because trials have only been carried out with adults.

As a parent of a child with a rare condition, you may have found out some information about off-label medicines that could help your child. If you think your child would benefit from an off-label medicine, take the information along to your child's specialist and discuss it with them.

ORPHAN DRUGS

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

OTHER TREATMENTS

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. Your child's specialists can advise on treatments to help manage symptoms. Condition support groups may also have useful information.

> *"When they said that my daughter had a rare condition that would affect her growth, I was stunned. I started to panic, wrongly assuming that she would never go to school, have a boyfriend, or get married."*
>
> **Parent carer**

RESEARCH & 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 have some information on clinical trials and questions to ask before getting involved. The healthtalk.org website has interviews with parents talking about how they found out about trials, why they decided to take part, or why they did not take part (see page 31 for contact details).

You can also find information about ongoing clinical trials on the NHS Choices website, or search on the Orphanet website (see page 31). Some support groups have information about ongoing research. It is worth asking the relevant condition support group about this.

TREATMENT BEYOND THE NHS

It can be devastating to find out your child has a rare condition, especially if there are limited options for treatment. It's only natural to want to find the best treatment for them, but it's important to be aware of the pros and cons of what's on offer, especially with claims of miracle 'cures' you might find on the internet. It's important to discuss any treatment you may be considering with your child's specialist. Below we outline some treatment options you may come across, and highlight any pitfalls to be aware of.

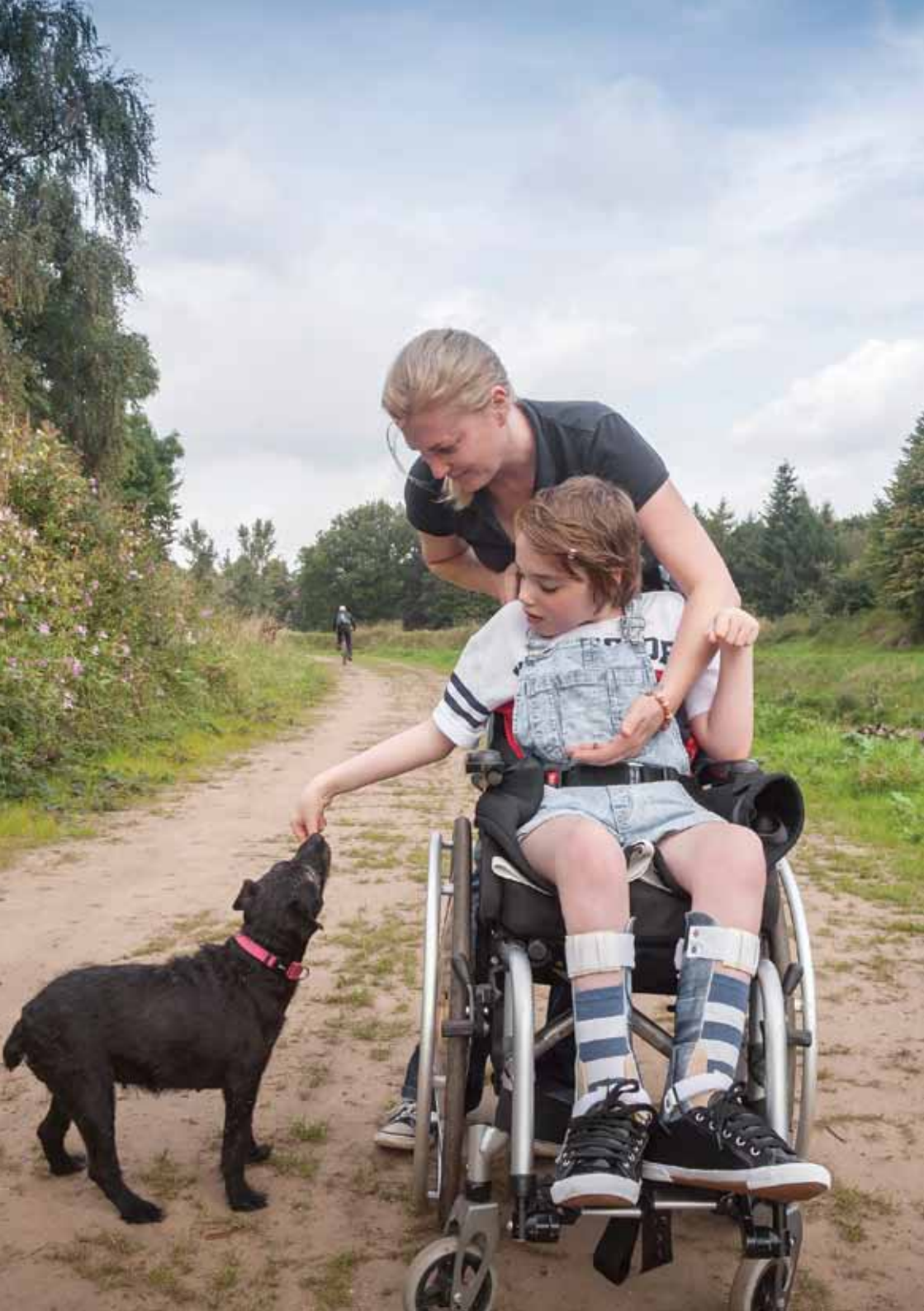
BUYING DRUGS AND TREATMENTS ONLINE

This may be very tempting, especially if a treatment claims to be successful and is not available in the UK. It's worth noting that the evidence for the treatment working may not be reliable. In addition, there is no guarantee that the drugs you are buying online are what they say they are because they don't have to undergo the same strict checks as prescribed medications in the UK. You should not change your child's diet or medication based on advice from any online sources. Treatments may not work and could even be harmful for your child.

EXPERIMENTAL TREATMENTS

Families might come across information about experimental treatments in their search for treatments for their child. It is important to discuss any information you come across with your specialist. 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 want your child to have an experimental treatment but professionals may not agree. This may be because they are 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 are rarely genuine and the cost of treatment can be very high.

TREATMENTS ABROAD

Some families may look into accessing specialist treatments abroad. This can often be very costly and the success of treatments can vary. In some cases, the NHS can fund treatment within Europe Economic Area countries (EEA). See the NHS Choices website for more details (page 31). 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 also get as much information as you can from a reliable, 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. It is important to discuss your intentions with your child's specialist in the UK as they may be able to offer some advice and may be involved in any follow-up treatment or care.

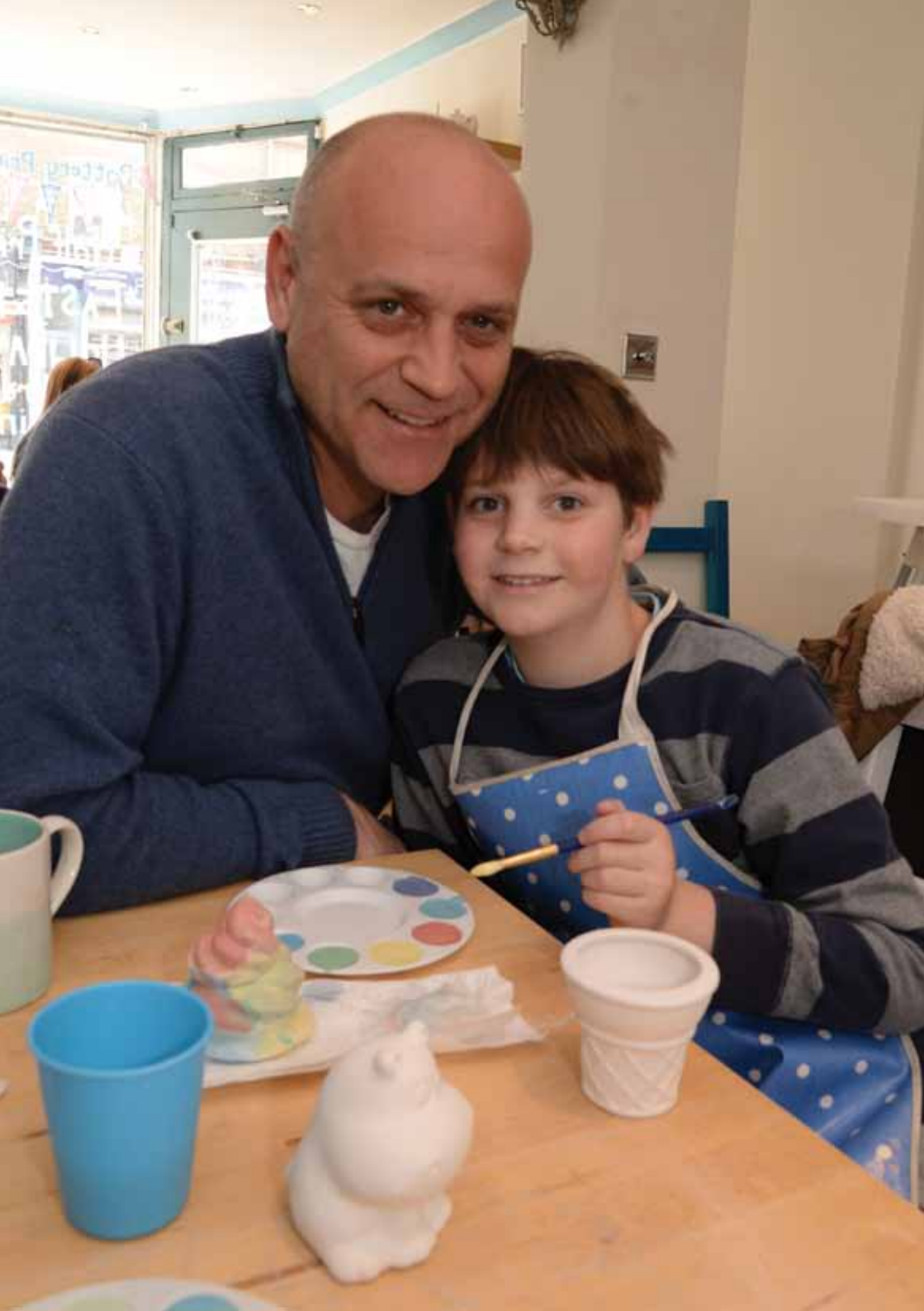
> *"Write down something positive about your child and remind yourself about it when you're having a bad day."*
>
> **Parent carer**

HELP WITH EDUCATION

Getting support for your child at nursery or in school may be more difficult if your child has a rare condition. You might find it helpful to ask your child's specialist or paediatrician to write a letter to the school listing your child's support needs. You may also want to take any reliable information you find on your child's condition for education professionals to read. Some rare condition support groups have leaflets for schools that you can find on their websites.

A child with additional needs is entitled to get help in an education setting based on their needs and not their diagnosis. The main point of contact for a parent whose child needs extra help in education is the early years or school's special educational needs coordinator (in England and Northern Ireland) additional learning needs coordinator (Wales) or additional support for learning assistant (Scotland). Tell the person you talk to what your worries are, give them examples and ask what support your child could have.

See our website for more information. Our Education Advice Service can also give you detailed information about your child's rights in education:
0808 808 3555 **www.contact.org.uk/education**



A PARENT'S STORY

When James was one I realised that he had something wrong. I just knew. Since that moment, we embarked on a journey that has been both heart-breaking and uplifting. It includes moments of darkness; when I was told how poorly he is, when he was misdiagnosed, when I was accused of abusing him, when he nearly died, and such great moments of happiness; when I see James laughing as he plays football with his friends, when I meet people who treat James in the same way as they treat others, when we partied at 10 Downing Street!

James has five rare blood conditions called Platelet Function Disorders (PFD). He also has Thrombocytopenia. This means his blood doesn't clot normally and if he knocks or bumps himself – which he does regularly – it could mean that he becomes seriously ill. James's condition is life-threatening but we live life with PFD, not running scared of it.

As PFDs are really rare, there is very little information about it. So I set up www.funnyblood.co.uk to help other parents find information more easily. It was also a way of doing something positive when there were so many negative things happening.

In fact, when I hear unhelpful remarks about Funny Blood I wonder what those people would do if they had a child with such a serious and rare medical condition? I guess I COULD just sit back and wait for life to happen? I CHOOSE not to. To quote Harper Lee:

'You never really understand a person until you consider things from his point of view... until you climb into his skin and walk around in it.'

So, we get on with life and grab as many opportunities as we can. In fact, that reminds me of another quote:

'Life's a journey, not a destination'

And this is OUR journey!

TOP TIPS FROM PARENTS

You may find the number of professionals and appointments you have to attend overwhelming. These tips from parents who have 'been there' may help.

- **Make a list of all the ways your child is affected** by their disability or condition to show doctors at appointments, or people in schools or nurseries.
- **Create a communication passport for your child** (see page 18). This can be taken to appointments and they are a quick and easy way of passing on information to the specialist about your child.
- **Get a diary or a big wall calendar** with space to mark appointment times.
- **Keep copies of all letters, appointments, and test results in a folder**. Keep a note of all the phone calls you make as well.
- **Prepare any questions you want to ask at an appointment beforehand.** Write these all down in an appointment notebook and take it along with you.
- **Take a friend or relative to your appointment to make notes while you talk.** It can be hard to remember what you are told, especially if you are upset.
- **Take a copy of any reliable information about your child's rare condition to local services** that may not be familiar with the condition, such as your GP, social worker or physiotherapist.
- **Work with health and other professionals to get the best help and support for your child**. Your expertise on their condition and the needs of your child is likely to be really important to this partnership.

- **Use a mixture of local services and specialised services that suit the needs of your family best** – this will reduce stress, time and expense by not making long unnecessary journeys. Make sure that you let professionals know this is your plan.
- **Don't be afraid to phone** if you feel that you need to see a specialist, or if you think you may have somehow got 'lost in the system'. 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. Support groups may also be a useful source of information about everyday issues.
- **Try to stick with the same professionals if possible**, so that your child gets a good level of care all the time from people that understand their needs.
- **As a parent you know your own child.** Trust your instincts and keep asking until you get answers.

> *"Finding others in similar situations, although their children don't have the same condition as my child, boosted my confidence. I feel as though my life has been saved from the despair and exclusion I felt."*
>
> **Parent carer**

USEFUL ORGANISATIONS

Children Living with Inherited Metabolic Diseases (Climb)

Climb is a UK organisation working 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. They also fund educational and primary research programmes and investigate treatments and medical services.

Helpline: 0845 241 2173
www.climb.org.uk

EURORDIS

EURORDIS is an alliance of patient organisations and individuals active in the field of rare diseases. Their website has information on rare diseases, orphan drugs, other patient experiences, training resources and news.

33 (1) 56 53 52 10
www.eurordis.org

Genetic Alliance UK

The Genetic Alliance UK is an umbrella body that represents many individual patient organisations. The Alliance 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 regional genetics centres.

0207 831 0883
www.geneticalliance.org.uk

Rare Disease UK

Rare Disease UK (RDUK) is the national campaign for people with rare diseases. It was established by the Genetic Alliance UK, the national charity of over 200 patient organisations supporting all those affected by genetic conditions. RDUK is working with health departments across the UK to implement the UK Strategy for Rare Diseases.

020 7831 0883
www.raredisease.org.uk

Unique

Unique is a source of information and support to families and individuals affected by any rare chromosome disorder and the professionals who work with them. Unique 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.

01883 723 256

www.rarechromo.co.uk

Healthtalk.org

Healthtalk.org shares more than 2,000 people's experiences of over 100 conditions and illnesses. In particular, they have a channel of parent stories from parents who have taken part in clinical research.

www.healthtalk.org

NHS Choices

NHS Choices has a wide range of health information including A-Z of conditions and a search function to check services near you like GPs, dentists and hospitals. It also holds information about clinical trials and a section on services, rights and paying for treatments abroad.

www.nhs.uk

Orphanet

Orphanet 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

National Organization for Rare Disorders (NORD)

Coalition of health organisations dedicated to helping people with rare conditions. They have a database of rare conditions and patient support organisations.

www.rarediseases.org

Contact has a Rare Conditions Information Officer who can help you find verified information about your child's rare condition and support if you haven't been able to find any. Call our helpline to access this free service **0808 808 3555** Please note we're unable to give medical advice.

GLOSSARY

Additional needs

This term 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 available to children or young people of the same age.

Clinical trials

A clinical trial is a particular type of clinical research that compares one treatment with another. It may involve patients or healthy people, or both. For example, a drug trial may test the safety of a drug in people and how well tolerated it is. Also the effectiveness of the drug may be compared to other available drugs.

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 their condition, medication, likes and dislikes and essential information for an emergency situation.

General Practitioner (GP)

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

Genetic condition

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

Gene replacement therapy

This is an experimental treatment where defective genes are replaced with healthy genes. These therapies are in the early stages of development and scientists and doctors are trying to get more information about how well they work. There are strict controls on these treatments and they will only be carried out in certain cases.

Metabolic disease

Metabolism is the process your body uses to get energy from the food you eat. A metabolic disease happens when abnormal chemical reactions in your body disrupt this process. When this happens, you might have too much of some substances, or too little of other ones that you need to stay healthy.

Off-label

A medicine becomes licensed for treating a condition once it has successfully undergone clinical trials. It can be difficult and expensive to run clinical trials for rare conditions. Doctors sometimes suggest prescribing 'off-label' medicines which they think might be beneficial, even though it has only been licensed to treat another condition or patient group. If your specialist is considering prescribing an unlicensed or off-label medication, they should inform you and discuss possible risks and benefits with you.

Occupational therapy

An occupational therapist 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.

Orphan drugs

Orphan drugs status was introduced by the European Union (EU) for the development of treatments for rare conditions. Usually, drug companies are not interested in developing a drug for so few patients, as the cost of development outweighs the profits they make. Orphan drug status comes with financial benefits for the drug companies as well as other assistance and support from the EU to develop the new medicines.

Paediatrician

A paediatrician is a doctor 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

A physiotherapist helps in the management and development of movement skills. Physiotherapy may include exercises to strengthen weak muscles and games to improve coordination and motor skills.

Specialist

A specialist doctor (or consultant) is a senior doctor who practises in one particular area of health, such as a cardiologist for the heart, or a neurologist for conditions affecting the brain. Once speciality training has been completed, doctors are able to apply for consultant posts. Consultants have ultimate responsibility for the care of patients referred to them.

Specialist clinics

A specialist clinic for a rare condition is a place where you can get information and support from many professionals under one roof to save numerous appointments. Often rare condition support groups work closely with the specialist centres for that condition.

Special educational needs

Children with special educational needs (SEN) have 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 coordinator

A special educational needs coordinator (SENCO) is a member of staff at a school or in early years' settings who has responsibility for coordinating SEN provision.

Speech and language therapy

Speech and language therapy helps children learn to communicate, either through speech or other methods. Speech and language therapists can also help if there are problems with swallowing.

Social worker

A social worker can help children and families by advising them on practical and financial issues, telling them about local services and sometimes helping to arrange support.

GET IN CONTACT

Our helpline advisers can support you with any issue about raising your disabled child: help in the early years, diagnosis, benefits, education and local support.

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We are Contact, the charity for families with disabled children.

We support families with the best possible guidance and information.

We bring families together to support each other.

We help families to campaign, volunteer and fundraise to improve life for themselves and others.

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