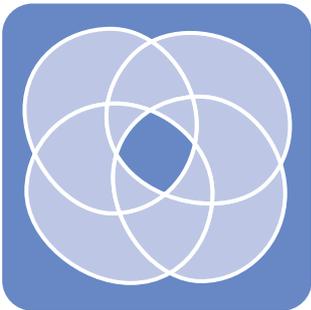
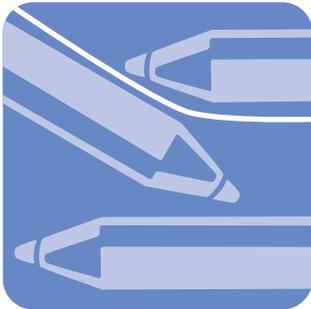
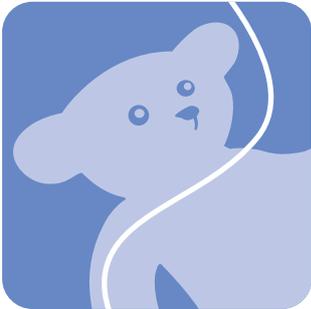


Information for parents

If your child has a rare condition



About this publication

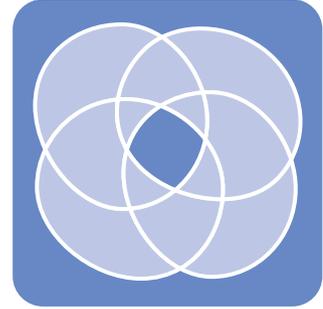
This is a guide for parents with young children who have recently been diagnosed with a rare condition, or who are in the process of getting a diagnosis.

It was developed in co-operation with families and this edition is an updated version based on the original information for parents booklet 'When your child has no diagnosis', adapted by kind permission of [Early Support](#), a UK government initiative. It was originally developed by [Early Support](#) in partnership with [Contact a Family](#), in response to requests from families, professional agencies and voluntary organisations for better standard information. Families were consulted about the content and the text reflects what parents who have 'been there before' say they would have liked to have known in the early days of finding out about their child's situation. This updated version of the publication was adjusted by the Early Support Wales team and [Contact a Family Wales](#) using feedback from parents and practitioners and in response to changes in the Welsh context.

To find out more about [Early Support](#) and to download copies of other [Early Support](#) materials, visit www.earlysupportwales.org.uk

Where a word or phrase appears in [colour](#), like this, it means either that it is a technical term that is explained in the text around it.

If the name of an organisation appears in [colour](#), it means that you can find their contact details at the back of the book.



Introduction

Discovering that your child has any condition can make you feel lost, confused, and isolated. If your child has a rare condition these feelings, particularly isolation can be magnified. Perhaps the doctor doesn't know very much about the condition or has never seen another affected child and can give you little advice about what you might expect over the coming months and years.

At present, it is estimated that there are between 5,000 and 8,000 distinct rare conditions. A **rare condition** is defined by the European Union as a disorder that affects fewer than five people in 10,000. Rare conditions can affect both children and adults and can occur at any time of life. Many conditions are chronic, progressive and disabling. Some are life threatening.

'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.' (Mother)

Getting information on rare conditions

Discovering that your child has a rare condition can cause a great deal of confusion. Sometimes it can take a long time to get an answer to your questions. Waiting for a name for the disorder and what it can mean for you as parents can be very frustrating.

Research has shown that there are several reasons why it is hard for you to find information on rare conditions. Some of these reasons are listed below:

Identifying the condition and ensuring the right diagnosis is made

In some cases, it can take longer to get a diagnosis for a rare condition. Some conditions have similar signs. The same conditions can affect individuals to varying degrees. Professionals are very keen to make sure that they give the right diagnosis and lessen the chance of giving incorrect information about a child's condition.

Lack of Information

For very rare conditions the research is often limited. It may be that very few medical professionals have seen a case of the condition and may be unfamiliar with it or unable to find information about the condition easily. This means it can take longer for the right tests to be arranged that can help determine your child's condition.

Referral to specialists

Some rare conditions are quite complex and children may need to be referred to different specialists to get as much information as possible. The more people that are involved in helping your child the longer it will take to make sure that everyone has the right information.

Even if you do not have a diagnosis you and your child are entitled to help and support. For more information, please call the [Contact a Family](#) helpline or consult the [Early Support Information for parents booklet 'When your child has no diagnosis'](#).

*"It really was not easy to find somewhere or someone who knew anything about a rare disorder or to have the time to give extra on a one-to-one basis in a busy playgroup."
(Parent)*



Making contact with other families

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

'Friends and relatives queued up to give me support but what I really wanted was to meet someone like me. When I approached my health visitor with this request she replied 'Oh no dear! There is no one else like you!' (Parent)

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

Many organisations recognise the benefits of linking people who have shared or will share similar experiences. Other parents can offer useful tips on the services that they have used and tips for coping with day-to-day tasks. [Contact a Family](#) offers a linking service (contact details can be found at the back of this booklet).

'We use a local group for special needs families, which is excellent.' (Father)

Many areas in the UK have a local parents' support group where families of children with all kinds of disabilities come together for mutual support and contact. It might be helpful to find out if there's one near you.

If your child has a rare condition it's unlikely that another child in the group would have the same disorder.

However, a lot of the issues you face on a day-to-day basis will be familiar to other parents and they often have practical advice to share.

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 that are available in your area and that they have already used.

How to find reliable medical information

Information for parents who have a child with a rare condition is available but can be difficult to find. With conditions that affect a relatively small number of individuals, little research is carried out and the information is often written in medical language.

Stories you see on the internet can be inaccurate or very extreme. It is always important to get information that has been verified as correct by a medical expert. [Contact a Family](#) produces the [Directory – the essential guide to medical conditions, disabilities and support](#) – which describes over 430 medical conditions and also includes details of support groups. The Directory can be viewed online or be purchased as a hard copy that is printed on a yearly basis. In addition, [Contact a Family](#) has produced a leaflet 'Finding quality medical Information on the internet'. All of these resources can be obtained by getting in touch with [Contact a Family](#) (contact details at the end of this booklet).

Even when you have found reliable information, it is important to remember that each child is different and a disorder will affect each child to varying degrees. It is always important to talk to the medical professional dealing with your child's case about your questions and concerns.

Often support groups can be a source of good understandable information.



Where to find out more

The UK-wide charity [Contact a Family](#) provides support, advice and information to families with disabled children, whatever the child's diagnosis – whether your child has acquired aplastic anaemia or Zellweger syndrome. [Contact a Family](#) has a base in England – their contact details are at the end of this booklet.

[Contact a Family](#) can:

- Put you in touch with support groups. They are in contact with many small national support groups, which cover many rare disorders
- Link you directly on a one-to-one basis with another family through their website www.makingcontact.org.uk, which is available in a range of community languages
- Put you in touch with other specialist voluntary organisations that may be able to help
- Give you medical information on all disorders affecting children and young people
- Provide free information guides on subjects that may be relevant – for example, [benefits, family relationships, and equipment, aids and adaptations](#)
- Provide leaflets for all family members, including fathers, mothers, grandparents and brothers and sisters
- Talk to you via an interpreter in over 100 languages if you prefer to use a language other than English/Welsh.

The [Contact a Family](#) website www.cafamily.org.uk has lots of useful information and includes an online [Directory – the essential guide to medical conditions, disabilities and support](#).

If you can't find a particular disorder listed in the directory, or do not have access to a computer, call the [Contact a Family helpline](#) (0808 8083555). Their advisers can go through information with you and will research a particular disorder for you if it's something they don't already hold information on. You can talk things through with a parent adviser, order free information guides, and find out about the support and help that is available in your area. The service is free and confidential.

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Other Voluntary Organisations

Climb (Children Living with Inherited Metabolic Diseases)

The United Kingdom's foremost provider of free metabolic disease information to young people, adults, families, professionals and other interested groups.

Free phone: 0800 6523181

Web: www.climb.org.uk

Email : info.svcs@climb.org.uk

Unique

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

Tel: 01833 330766

Web: www.rarechromo.co.uk

Email: info@rarechromo.org

About Early Support

Early Support in Wales is the Welsh Government's mechanism for achieving better co-ordinated, family-focused services for young disabled children and their families across Wales.

Early Support builds on existing good practice. It facilitates the achievement of objectives set by broader initiatives to integrate services, in partnership with families who use services and the many agencies that provide services for young children.

To find out more about **Early Support** and associated training opportunities and to view or download other resources produced by the programme, visit www.earlysupportwales.org.uk

This information resource is one in a series produced in response to requests from families, professional agencies and voluntary organisations for better standard information about particular conditions or disabilities. The other titles in the series are:

- Autistic spectrum disorders and related conditions
- Behaviour
- Cerebral palsy
- Deafness
- Down Syndrome
- Learning Disabilities
- Multisensory impairment
- Neurological disorders
- Sleep
- Speech, language and communication needs
- Visual impairment
- When your child has no diagnosis

Other Early Support information about services is available separately. To download any of the **Early Support** materials mentioned here, see www.earlysupportwales.org.uk

Information for parents
If your child has a rare condition

Contact a Family Cymru provides support, advice and information for families with disabled children, no matter what their disorder or disability. **Contact a Family** provides advice about financial and practical help as well as information on medical disorders and disabilities. They also put families in touch with others through support groups and one to one linking, assist parents to develop their own support groups and provide a voice to raise awareness and campaign for families.

The **Contact a Family** website contains all publications, also available in paper format, including the Contact a Family Directory.

Contact a Family Cymru
33-35 Cathedral Road
Cardiff
CF11 9HB

Helpline: 0808 808 3555

Tel: 029 2039 6624

Web: www.cafamily.org.uk

Email: wales.office@cafamily.org.uk

Copies of this resource can be
downloaded from
www.earlysupportwales.org.uk

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Llywodraeth Cymru
Welsh Government