



About diagnosis: Rare disorders

Information about rare disorders and the help available

- Rare disorders
- Support from others
- Finding out more

Learning that your child has any condition can trigger a range of difficult feelings such as loss, confusion and isolation. When a condition is rare you and your family may experience the added challenges of getting a diagnosis, finding clear information and advice, and treatment options may vary depending on where you live.

What is a rare disorder?

There are over 5,000 known rare disorders. More disorders are discovered as medical science advances. They affect both children and adults and can occur at any time of life. The European Union's definition of a rare disorder is 'a condition which affects five or less people in every 10,000'. In the UK, it is estimated that out of the 15,000 disabled children born each year, 1,200 children will have a rare disorder.

Rare disorders and diagnosis

Discovering that your child has a rare disorder can cause a great deal of confusion. It can take time to get answers to your questions. Waiting for a 'name' for the disorder and what this means for you as parents can be very frustrating.

Research has shown that there may be delays in getting information about rare disorders and we have listed some of the reasons for this:

■ Identifying the condition and ensuring the right 'diagnosis' is made

Many rare disorders share similar signs. Conditions can also affect individuals in different ways and to varying degrees. So professionals are keen to make sure they offer accurate information and lessen the chance of giving incorrect advice about a child's condition. This means it can take longer before they can give you the information you need or a name for your child's condition.

■ Lack of information

With very rare conditions, research is often limited. The medical professional involved with the care of your child may not be familiar with the condition or able to find information on it easily. This means it can take longer for the right tests to be arranged that might identify the name of the condition affecting your child.

■ Referral to specialists

Some rare disorders are quite complex and children may need to be referred to different specialists to get as much information as possible. Of course, the more people involved in helping you and your child, the longer it will take to ensure that everyone has all the right information.

There is often help available for your child before the results of any tests and discussions are confirmed. You will almost certainly be entitled to help and support at this time too. For further information, please call the Contact a Family helpline.

Making contact with others affected by rare disorders

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. While this is occasionally true, it may be that the professional concerned has just not seen a child with that condition, or even heard of it.

“Friends and relatives queued up to offer me a shoulder to cry on 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’s no one else like you!’”

Parent

However, there are often other families in the country affected by the same condition. With the internet and support groups making international contacts, it may be that you can get in touch with others in the same situation.

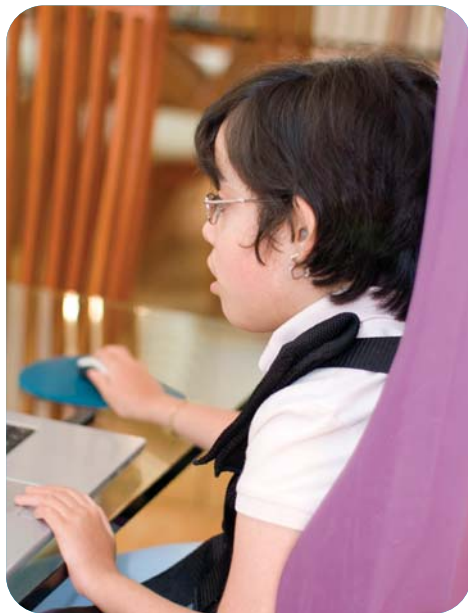
“With help from Contact a Family we have found sharing our experiences with other families in the same sort of situation, especially chatting through email, has helped us to cope.”

Parent

Finding out more about your child’s rare condition

Information for parents whose children have rare conditions is available but can be very hard to find. With conditions that affect a relatively small number of people, little research is done and the information that is available is written in very medical language. Support groups can be a good source of understandable information. The Contact a Family Directory has descriptions of many rare disorders and our helpline can help to find more information. The Contact a Family Directory is available online at Web: <http://www.cafamily.org.uk/medicalinformation>

For more guidance on how to search for reliable and accurate information, please read our guide ‘Sources of medical information’, part 9 in this series.



How Contact a Family can help

Contact a Family can help you to find other families with similar experiences who have children affected by the same condition as your child through extensive UK contacts and growing communities worldwide. We have information on support groups for many rare conditions, and you could link with other families through our internet based linking scheme, Web: <http://www.makingcontact.org>. You can also find us on Facebook.

For information on rare disorders and help to contact other families, please call the Contact a Family Helpline.

Other guides in this series

- 1 How we can help
- 2 Support for parents
- 3 What is developmental delay?
- 4 Understanding behaviour
- 5 Getting a diagnosis
- 6 What if we don't get a diagnosis?
- 7 What is a genetic condition?
- 8 Rare disorders
- 9 Sources of medical information
- 10 Concerned about your child
- 11 Practical and financial help

Getting in contact with us

Free helpline: **0808 808 3555**
Open Mon–Fri, 10am–4pm;
Mon, 5.30–7.30pm

www.cafamily.org.uk
www.makingcontact.org

Contact a Family Head Office:
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Textphone 0808 808 3556
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Other information available

- NHS and caring for a disabled child (England & Wales)
- Disabled children's services (England & Wales/Scotland)
- A guide to claiming DLA for children (UK)
- Finding medical information on the internet (UK)
- Understanding your child's behaviour (UK)
- Relationships and caring for a disabled child (UK)

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