

# LIVING WITHOUT A DIAGNOSIS

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INFORMATION FOR PARENTS OF DISABLED CHILDREN



contact

*“The way I feel about  
her is pride; she is  
special.”*

**Parent carer**

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## INFORMATION IN THIS GUIDE

Please look in the glossary at the back of this guide to find an explanation of any unfamiliar words.



# INTRODUCTION

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This guide is for parents and carers whose child has a learning difficulty or health problem, but do not know its cause. It contains information on:

- ***why it can be hard to get a diagnosis***
- ***who can help you***
- ***what help and support you can expect***
- ***meeting other parents***
- ***top tips.***

When trying to get a diagnosis for their child, families can go through a range of emotions. We have number of guides with topics that may be useful to parents in this situation. 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.***

# ABOUT DIAGNOSIS

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Once you realise your child may have a learning difficulty or health problem, getting a diagnosis may take some time.

The Genetic Alliance UK estimates that about 50 per cent of children with a learning disability have no definitive diagnosis. There are some children who are disabled, but nobody is able to explain the cause.

Some families may be told that their child has global developmental delay (GDD), if they are slow to reach two or more milestones in all areas of their development.

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See our guide ***Developmental Delay*** for more information. It's free from our helpline: **0808 808 3555** or from **[www.contact.org.uk/dev-delay-guide](http://www.contact.org.uk/dev-delay-guide)**

## WHY MAKING A DIAGNOSIS CAN BE DIFFICULT

Because of improvements in science and medicine, people sometimes believe that doctors can always diagnose a child's condition. Sometimes this isn't the case and this can be disappointing. There are a number of reasons why making a diagnosis can be difficult:

- ***There are more than 6,000 known rare conditions.*** Doctors rarely see children with these conditions, making it harder to recognise them
- ***Many conditions have similar features or symptoms,*** which makes it more difficult for doctors to make a diagnosis
- ***Some children have a number of features or symptoms that do not fit into one specific condition***
- ***Each condition can affect children in different ways***
- ***Certain symptoms associated with a condition may not appear until your child is older*** – this may result in a late diagnosis, or even a change in diagnosis.

## WHY DID IT HAPPEN?

Some parents worry that it is their fault their child is disabled because of something that happened before the baby was conceived, or while the mother was pregnant. It is important to remember that it is rarely anyone's fault. There are many reasons why children are born with a disability or medical condition, one of the most common being changes in genes (our body's blueprint) or chromosomes (the structures that DNA is strung onto). These are called genetic conditions. Other reasons may be:

- ***being born too early (or prematurely)***
- ***environmental problems, such as substances the baby is exposed to while the mother is pregnant (for example, lead or pesticides)***
- ***difficulties during pregnancy, such as infections in the mother (for example rubella)***
- ***asphyxia, a lack of oxygen or trauma, during, or shortly after birth.***

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

**Parent carer**

## WHO CAN HELP?

You can discuss your concerns about your child's development with your health visitor. You can also discuss your concerns about your child with your general practitioner (GP).

Your GP may refer your child to a paediatrician, someone who specialises in conditions affecting children and young people. They in turn may refer your child to a consultant who specialises in a particular area of medicine. If your child has lots of symptoms, they might be referred to different specialists to see if any of them can make a diagnosis.

Other health professionals may be involved in assessing your child and how well they can cope with certain activities. For example, speech and language therapists (SALTs), physiotherapists and occupational therapists (OTs).



Find out more about the different professionals who can help in our guide **Concerned about your child**, available free from our helpline or to download at: [www.contact.org.uk/parent-guide-concerned](http://www.contact.org.uk/parent-guide-concerned)

*"Be careful with the internet, there's so much out there and it's so easy to get poor information and frighten yourself."*

**Parent carer**



## TOP TIPS

- **It's never too early to ask for advice.** If you have worries about your child, talk to your family doctor (GP), **health visitor** or nurse. They might suggest your child sees a **paediatrician** or other specialists, who will try to work out why your child is finding things hard.
- **Make a list** of all the things that worry you about your child. You can show this to professionals when you see them.
- **Keep a record of your child's progress.** Take photos and keep a diary.
- **Write down any questions** you want to ask before you have an appointment.
- **Keep copies of all the letters** and results you get in a folder.



# TESTS, ASSESSMENTS AND MEDICAL APPOINTMENTS

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Trying to get a diagnosis can mean your child has lots of different tests, assessments and appointments. This can be a worrying and anxious process.

## WHAT CAN YOU DO?

As a parent you may suspect a particular type of condition, so you could ask for a referral to a specialist service (for example, for metabolic conditions – see page 25). If you want to do this, it can be helpful to take along to the appointment a list of all the ways your child is affected to explain why you think a referral to a specialist is needed. Support groups for the condition you have in mind may be able to help you find information and the right specialists to make a diagnosis.

If you think your child could benefit from a particular assessment or test then discuss this with your medical professional. If you find reliable evidence to support this, bring a copy along to your appointment to show the doctor.

### FINDING INFORMATION TO HELP YOUR CHILD

National condition support groups may have information that's useful to you if your child has certain symptoms, even if they don't have a named diagnosis. For example, the National Autistic Society may be able to offer useful resources for helping with behaviour, ICAN and The Communication Trust have information to help with speech and language and SCOPE can help provide solutions to issues with poor muscle control.

## 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 when 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.*

## **NEW CONDITIONS ARE BEING IDENTIFIED ALL THE TIME**

Doctors are now able to diagnose more conditions than ever before because of advances in medical knowledge and improvements in testing techniques and equipment.

If it is suspected that the cause of your child's condition is genetic, then you may be referred to a genetics service, which are based at regional genetics centres. Ask your GP or paediatrician for a referral to a genetics centre if you think your child has a genetic problem.

### **SWAN UK (Syndromes Without A Name)**

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SWAN is a support network providing information and support to families of children with undiagnosed genetic conditions. They offer an online forum, family fun days and support and information in hospital, at home and in your local communities. It is run by the charity Genetic Alliance UK (see page 23).

**[www.undiagnosed.org.uk](http://www.undiagnosed.org.uk)**



We have more information about genetic conditions, including a short podcast about what to expect when visiting a genetics service, at

**[www.contact.org.uk/genetics](http://www.contact.org.uk/genetics)**

## WHAT TO DO IF YOU ARE NOT HAPPY

If you feel strongly that health professionals have not explored all ways of getting a diagnosis for your child, you might be able to get a second opinion.

For example, 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.

If after seeing a specialist doctor you would like a second opinion, you need to 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 and explain the reason you are asking for the referral.

### HOW IMPORTANT IS GETTING A DIAGNOSIS?

While most parents say they would like a diagnosis, over time some find that getting a 'name' is not as important as it was at first. For these parents, the most important thing is ensuring that their child's day-to-day needs are met.

Others, however, want a diagnosis to help make sure their child has the best possible support and treatment. A lot of parents fear that their child will not be able to get the support they need without a firm diagnosis.

Your child is entitled to support for their needs whether they have a diagnosis or not. The support they receive should be based on their additional needs and not the name of a condition. See the next section information about this.

*"I felt that getting a name for our daughter's condition would help us when explaining to others what was wrong, but in the end, it didn't matter. All the professionals involved needed to know was that the immediate difficulties were, so the right care could be arranged."*

**Parent carer**



# GETTING THE RIGHT SUPPORT

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Below we describe the support that your child and you as a carer/family are entitled to, whether you have a diagnosis or not.

## COMMUNICATING YOUR CHILD'S NEEDS

iMake a list of all the ways your child is affected and take copies along to all medical appointments and assessments.

### Communication passports

You might find it helpful to create a communication passport for your child. 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.

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 CALL Scotland have templates for communication passports on their website. Visit [www.communicationpassports.org.uk](http://www.communicationpassports.org.uk)

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 Mencap also have a hospital passport for children with a learning disability on their website: [www.mencap.org.uk/advice-and-support/health/our-health-guides](http://www.mencap.org.uk/advice-and-support/health/our-health-guides)

## TREATMENT AND SUPPORT STRATEGIES

Treatment, therapy or other support services should be tailored to your child's needs, not the name of their condition. Strategies to help your child's mobility or speech and language development are suitable for many children with different conditions.

## SERVICES AND SUPPORT FROM YOUR LOCAL AUTHORITY

Social services departments of local authorities are legally responsible for arranging support for disabled children, their siblings, and their carers. You are entitled to have a social services assessment of your child's needs, and your needs as a parent carer, whether your child has a diagnosis or not.

An assessment is the start of the process to decide if services are needed. It is an opportunity for you to tell a professional about your child and family's needs. An assessment is important because it can lead to a number of services being provided, like practical help in the home and short breaks.

You can request an assessment by contacting your social services department (in Scotland this is known as the Social Work Department, and in Northern Ireland it is the Health and Social Services Trust). Your GP, paediatrician or health visitor can also ask on your behalf.



Our website has more information about accessing services, plus in-depth guides and factsheets at: [www.contact.org.uk/social-care](http://www.contact.org.uk/social-care) or you can call our helpline: **0808 808 3555**.

## GETTING FINANCIAL HELP/SUPPORT

You might be entitled to receive benefits such as Disability Living Allowance, depending on the level of support or care that your child needs. Our freephone helpline offers expert confidential advice on benefits, and we have a list of charitable grants parents can apply to for help.

Parents often worry that when they fill out forms to get services or financial assistance, having no diagnosis for their child will work against them. You can ask your specialist or paediatrician to write a letter to outline the difficulties and/or symptoms your child experiences and what



effect this has on daily life to support your claims. We can offer advice to families on financial support regardless of whether your child has a diagnosis.



See our guide to **Money Matters**, a checklist of financial help you may be entitled to at [www.contact.org.uk/finance](http://www.contact.org.uk/finance) or contact our helpline and ask for your free copy: **0808 808 3555** [helpline@contact.org.uk](mailto:helpline@contact.org.uk)

## GETTING SUPPORT FOR YOUR CHILD'S EDUCATION

Your child is entitled to have extra support to help them at their nursery, pre-school or school, if they need it. This does not depend on having a diagnosis.

However, getting support for your child at nursery or in school may be more difficult if you do not have a named condition. You can improve the setting's/school's understanding by getting a letter written by the child's specialist or paediatrician to list their needs and how best they can be supported.

A child with additional learning needs is entitled to get help in the education setting based on their needs, and not their diagnosis.

The main point of contact is the early years or school's special educational needs coordinator (England and Northern Ireland), Additional Learning Needs Co-ordinator (Wales) or Additional Support for Learning Assistant (Scotland).



More on extra help in education is at [www.contact.org.uk/education](http://www.contact.org.uk/education)  
Our helpline's Education Advice Service can also support you:  
**0808 808 3555** [helpline@contact.org.uk](mailto:helpline@contact.org.uk)

# MEETING OTHER PARENTS

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Parents often say that what helped them most was meeting other parents who are in, or have gone through, a similar situation. Meeting other parents can be a chance to hear useful tips, find out about local services and learn strategies for dealing with common issues like sleep and behaviour.

## LOCAL PARENT SUPPORT GROUPS

Many areas in the UK have local parents' support groups where families with children with all kinds of disabilities meet up. Local groups have the advantage of meeting regularly and provide a support network near your home if you need one.



Visit our website or call our helpline to find groups in your area:

**0808 808 3555**    [www.contact.org.uk/parent-support-groups](http://www.contact.org.uk/parent-support-groups)

## OUR ONLINE COMMUNITY

Our online community is a safe and trusted space where you can talk to other parents about anything and everything that affects you and your child – from medical conditions to healthcare, holidays and higher education.



Register to talk to parents, or log in with your Facebook account:

[www.contact.org.uk/community](http://www.contact.org.uk/community)



## TOP TIPS FROM PARENTS

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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 15). This can be taken to appointments and it is 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 you can use 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.

*“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 carer**

## A PARENT'S STORY

Daniel, now 23, is the second of three boys. The main problem of not having a diagnosis was not knowing the cause of Daniel's severe developmental delays. We knew from the age of three, after an MRI at Great Ormond Street Hospital, that he has hypoplasia of the corpus callosum (underdevelopment of the middle structure of the brain). But this didn't explain all his difficulties.

Daniel is very delayed, we were told he was between 9 and 15 months in development. He is non-verbal and is incontinent. Of course, we didn't know any other parents with a child quite like Daniel, but we had friends with other diagnosed children online. We had read that 60% of children never receive a diagnosis, so we just imagined he was one of the ones who would never have a diagnosis.

When we moved to Holland, we thought we should get him known at the local teaching

hospital in case he should develop seizures again.

The genetics department at the hospital tested him for Angelman syndrome, (Daniel's fourth Angelman test!), along with male Rett syndrome. Following this, a few years later they thought of him as a possible candidate for a newly developed test for the extremely rare Pitt Hopkins syndrome.

Then, completely out of the blue, we received the call from our geneticist informing us that our almost 16-year old finally had a diagnosis: Pitt Hopkins syndrome. We'd never heard of it before, and we certainly didn't know anything about it – even the name was a mouthful!

We later found out that there were less than 16 confirmed cases worldwide. Our immediate thought was to look for a support group, but there was nothing available – so this is where the story of the Pitt Hopkins Syndrome support group starts!

# USEFUL ORGANISATIONS

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## **The Communication Trust**

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The Communication Trust is a campaigning voice for children with speech, language and communication needs. The Trust also has a number of useful resources.

**0207 843 2526**

**[www.thecommunication.trust.org.uk](http://www.thecommunication.trust.org.uk)**

## **ICAN**

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ICAN supports children with speech, language and communication difficulties. They offer a range of information services that provide help and advice to parents and practitioners about speech, language and communication.

**0207 843 2544**

**[www.ican.org.uk](http://www.ican.org.uk)**

## **Genetic Alliance UK**

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Genetic Alliance UK aims to improve the lives of people affected by genetic conditions. The Alliance runs several projects, including SWAN UK (see page 12), and has information on genetic testing and services, including a list of UK regional genetics centres.

**0207 831 0883**

**[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)**

## **Mencap**

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Mencap work to improve the lives of children and adults with a learning disability and their families. They run various projects and programmes for people with a learning disability, their families and professionals, including local groups and sport.

**Helpline: 0808 808 1111**

**[www.mencap.org.uk](http://www.mencap.org.uk)**

## **National Autistic Society**

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National Autistic Society (NAS) is a UK-wide charity for people who have autism (including Asperger's syndrome) and their families. NAS provides information, support and services. Even though your child may not have been diagnosed with autism, NAS has useful information.

**[www.autism.org.uk](http://www.autism.org.uk)**

## **SCOPE**

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SCOPE supports disabled people and their families in the UK by providing help and information, services and campaigning. The charity specialises in helping people with cerebral palsy but will support any disability.

**Helpline: 0808 800 3333**

**[www.scope.org.uk](http://www.scope.org.uk)**

# GLOSSARY

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## **Additional learning needs coordinator**

The person at a school or in early years' settings who is responsible for coordinating extra help in education in Wales.

## **Additional needs**

Any additional support, beyond services available to all children of the same age, that a child or young person who has a condition, difficulty, or challenge, needs.

## **Additional support for learning assistant**

The person at a school or in early years' settings who is responsible for coordinating extra help in education in Scotland.

## **Asphyxia**

A condition where there is a lack of oxygen in the blood and a build up of carbon dioxide, usually due to stopping breathing or suffocation.

## **Communication or hospital passport**

A communication or hospital passport is a booklet that is used to pass on crucial details about a child or young person with additional support needs. It contains information about their condition, medication, likes and dislikes and essential contacts in an emergency situation.

## **Disability Living Allowance**

Disability Living Allowance (DLA) is the main benefit for children who have a condition or long-term illness. To make a successful claim for DLA, you have to show that your child needs substantially more care, attention or supervision than other children of the same age who don't have disability or health condition. **Warning!** You should not claim DLA for a child who is subject to immigration control without first speaking to a specialist adviser, as a claim could affect your right to stay in the UK.

## **General practitioner (GP)**

Your GP will be the first person to ask for medical help and advice. They can ask for your child to be seen by a specialist.



## Genetic condition

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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 through the parents' genes, but others are caused by changes to DNA and chromosomes.

## Global developmental delay

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A child may be described as having global developmental delay (GDD) if they have not reached two or more milestones in all areas of development (called developmental domains). These areas are:

- ***motor skills – gross motor skills (sitting up or rolling over) and fine motor skills (picking up small objects)***
- ***speech and language – babbling, imitating speech and identifying sounds***
- ***cognitive skills – the ability to learn new things, process information and/or organise thoughts***
- ***social and emotional skills – interacting with others and development of personal traits and feeling.***

Our guide **Developmental delay** has more information. Call our freephone helpline on **0808 808 3555** for a free copy.

## Genetics service

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Genetics centres in the UK are organised on a regional basis and include both clinics, where you may see a clinical geneticist or a genetics counsellor, and laboratory services, where tests are carried out. Genetics services may have other professionals based within them. The service will support anybody who is affected by a genetic condition, or if it is suspected that they have a genetic condition. The Genetic Alliance can let you know where your nearest your nearest genetic centre is (see page 23).

## Metabolic condition

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Metabolism is the process your body uses to make energy from the food you eat. A metabolic condition occurs when abnormal chemical reactions in your body disrupt this process. When this happens, you might have too many of some substances, or too few of others that you need to stay healthy.

## Health visitor

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A health visitor is a nurse who has undertaken extra training to work with families in the community. They can give practical advice on day-to-day matters such as feeding, sleep, and behaviour. Some health visitors have specialist roles, such as working with children with disabilities.

## Occupational therapist

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An occupational therapist pays particular attention to hand-to-eye coordination, perception and manipulative skills. They can give advice and sometimes provide suitable aids for feeding, dressing, toileting, bathing, play and literacy skills.

## Paediatrician

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A paediatrician is a doctor who specialises in looking after babies, children and young people. A paediatrician can coordinate the work of services involved in the management, care and education of a child.

## Physiotherapists

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

## Prematurity

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A 'premature' or 'preterm' baby is one that is born after 24 and before 37 full weeks after the first day of the mother's last period.

## Rare condition

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The European Union says a condition is considered to be rare if it affects fewer than five people in every 10,000. Rare conditions 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.

## Social services assessment

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As a parent, you have the right to ask for your child's needs to be assessed by social services. In Scotland this is known as the Social Work Department. In Northern Ireland it is called the Health and Social Services Trust. You also have the right to ask for an assessment for yourself. An assessment could lead to services for your child being provided, or services begin offered to help you as their carer. You also have a right to ask for a carers assessment. An assessment could lead to services, or direct payments to arrange services or support for you and your child.

## **Specialist**

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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.

## **Special educational needs (SEN)**

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

## **Special educational needs coordinator (SENCO)**

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The person at a school or in early years' settings who is responsible for coordinating extra help in education in England.

## **Speech and language therapist (SALT)**


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A speech and language therapist helps children learn to communicate, either through speech or other methods. They can also help if there are problems with eating, drinking and swallowing.

# GET IN CONTACT

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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.

 0808 808 3555

 [info@contact.org.uk](mailto:info@contact.org.uk)

 [www.contact.org.uk](http://www.contact.org.uk)

 [twitter.com/contactfamilies](https://twitter.com/contactfamilies)

 [facebook.com/contactfamilies](https://facebook.com/contactfamilies)

 [youtube.com/contactfamilies](https://youtube.com/contactfamilies)

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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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