



Living without a diagnosis

Information for families

Introduction

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.

About this series of guides

Around the time of diagnosis, families can go through a range of emotions. The 'About Diagnosis' series by Contact a Family deals 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. The other guides currently available in our 'About Diagnosis' series are:

- *Developmental delay*
- *Living with a rare condition*

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Colour key to reading this guide:

Words in **blue** in this guide are organisations listed in 'Contacts' from p10. Words in **brown** in this guide are explained in the 'Glossary' on page 12. The colour key will not work if you print the guide out in black and white.

Living without a diagnosis

Once you realise your child may have a learning difficulty or health problem, getting a diagnosis might 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 with a disability, but nobody is able to explain the cause.

Some families may be told that their child has global developmental delay (GDD). Find out more in our 'About Diagnosis' guide, *Developmental delay*.

Why did it happen?

There are many reasons why children may be born with a disability or medical condition, including:

- difficulties during pregnancy, such as infections in the mother (for example rubella)
- being born too early (**prematurity**)
- environmental problems, such as substances the baby is exposed to while the mother is pregnant (for example, alcohol, lead or pesticides)
- **genetic conditions** caused by changes (mutations) in genes (our body's blue-print) or chromosomes (the structures that DNA is strung onto).
- **asphyxia**, a lack of oxygen or trauma, during, or shortly after birth.

Parents may worry that it may be their fault their child has a disability, 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.

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www.cafamily.org.uk

Why making a diagnosis can be difficult

Because of improvements in science and medicine, people may 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 can 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

Who can help?

Many parents first discuss their concerns about their child's development with their **health visitor**. Families may also discuss their concerns about their child with their **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 can they cope with certain activities. For example, **speech and language therapists (SALTs)**, **physiotherapists** and **occupational therapists (OTs)**. If you would like to know more about what these professionals do you can see Contact a Family's guide *Concerned about your child*.

Tests, assessments and medical appointments

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



What can parents 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 disorders**). If you want to do this, it can be helpful to take a list of all the ways your child is affected along to the appointment to explain why you think a referral to a specialist is needed.

If you think your child may 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.

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 is based at a regional genetics centre. Often children with a genetic condition will have distinctive facial features (also called dysmorphic features).

Ask your GP or **paediatrician** for a referral to a genetics centre if you think your child has a genetic problem, or if you think your child has dysmorphic features. For more information, see contact details for the [Genetic Alliance UK](#) on page 9 of this guide.



What to do if you are not happy

If you feel strongly that all ways of getting a diagnosis for your child have not been explored, you may be able to get 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 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.

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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 a Family can also offer support and advice if you are unhappy with your child's care.

How important is getting a diagnosis?

Whilst most parents say that they would like a diagnosis, over time some parents may 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.

Parents may, however, want a diagnosis to help make sure their child has the best possible support and treatment.

Getting the right support

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 get support for their needs whether they have a diagnosis or not.** In practice, though, you may find it harder to get support for your child if they do not have a diagnosis. See the list below for what your child and you as a carer/family should be entitled to.

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



- You are entitled to have a **social services assessment** (in Scotland this is known as the Social Work Department, and in Northern Ireland it is the Health and Social Services Trust), of your child's needs and your needs as a parent carer, whether your child has a diagnosis or not. You can request an assessment by contacting your social services department. Your GP, paediatrician or health visitor may also ask on your behalf.
- You may 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.
- 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 knowing the cause of their diagnosis. Contact a Family has information on **special educational needs** (SEN) and can also give advice through their helpline.

Communicating your child's needs

Repeatedly having to explain your child's medical problems to different professionals can be frustrating. You can make a list of all the ways your child is affected and take copies along to all medical appointments and assessments. You can also keep a record of your child's progress using the [Early Support Development Journal](#).

Many parents find it helpful to create a **communication passport** for their child to pass on crucial details, such as information about their **additional needs**, medications, likes and dislikes and what to do in case of emergency. For example, the [Foundation for People with Learning Disabilities](#) have a template you might use.

Getting financial help/support

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. Contact a Family's freephone helpline can offer advice to families on financial support regardless of whether your child has a diagnosis.

Getting support for your child's education

Getting support for your child at nursery or in school may be more difficult if you do not have a named condition. Parents can improve the settings/school's understanding by getting a letter written

"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 what the immediate difficulties were, so the right care could be arranged." Parent

by the child's specialist or paediatrician to list the child's 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 for a parent carer whose child has special educational needs, (or additional learning needs in Wales), is the early years or school's **special educational needs coordinator** (SENCO). To find out more, call Contact a Family's freephone helpline on 0808 808 3555.

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www.cafamily.org.uk

Meeting other parents

Parents often say what helped them most was meeting other parents who are in, or have gone through, a similar situation. These parents may have useful tips about using local services and useful strategies for dealing with common issues like sleep and behaviour with their children.

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. Call Contact a Family's freephone helpline for contact details of groups in your area.

National condition support groups

It is worth remembering that whilst your child may not have a named diagnosis, many organisations have information that might be useful to you. 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.

How Contact a Family can help you

We are a UK-wide charity providing advice, support and information to families with a child with additional needs or a rare medical condition in the UK. Call our freephone helpline on 0808 808 3555. We also have access to interpreters in more than 170 languages.

We provide medical information on many conditions, including rare conditions. See www.cafamily.org.uk/medical-information or call our freephone helpline.

We can put you in touch with condition specific support groups and local support groups and other families through our online family linking scheme – MakingContact.org at www.makingcontact.org

Where there is no UK support group, we offer a one-to-one family linking scheme for rare conditions, where possible.

We also support groups of parents and carers that set up and run local and national support groups.

"I'm tired of having to explain to people that my child isn't naughty but has an unnamed condition."

Parent

Useful organisations

Bristol Children's Hospital

Tel: 0117 923 0000

Bristol Children's Hospital has a good template for a hospital passport, which can also be used in other settings. Visit the website www.uhbristol.nhs.uk and search for 'passport'.

Early Support

Tel: 0207 843 6000

Email: earlysupport@ncb.org.uk

Web: www.councilfordisabledchildren.org.uk/earlysupport

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

Early Support has lots of information on their website, including guides on behaviour and sleep. They also produce the Early Support Development Journal.

Foundation for People with Learning Disabilities

Web: www.learningdisabilities.org.uk

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



Genetic Alliance UK

Tel: 0207 704 3141

Email: contactus@geneticalliance.org.uk

Web: www.geneticalliance.org.uk

Genetic Alliance UK is an umbrella body that represents many patient organisations. They aim to improve the lives of people affected by genetic conditions. The Alliance runs several projects, including [SWAN UK](#) and has lots of information on genetic testing and services, including a list of all UK regional genetics centres.

ICAN

Information line: 0845 225 4073

Email: info@ican.org.uk

Web: www.ican.org.uk

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.

(continues on page 11)

Freephone helpline: **0808 808 3555**
www.cafamily.org.uk

Top tips

Parents can find the number of appointments and professionals they meet overwhelming. These tips from parents who have 'been there' can help you to manage your diagnosis journey.

- Make a list of all your concerns about your child. This can be useful to show doctors at appointments, or people in schools or nurseries.
- Keep a record of your child's progress. Keep a diary, take photos and videos if possible. As well as being a useful record to show to doctors, it may also be comforting to look back and see the progress your child has made.
- Create a communication passport for your child. This passport can be taken to appointments and they are a quick and easy way of passing on information to professionals about your child.
- Get a diary or a big wall calendar 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.
- Write down the questions you want to ask at an appointment beforehand.
- Take a friend or relative along to your appointment so they can make notes while you talk. It can be hard to remember everything you are told.
- Do not be afraid to ask your specialist questions that are concerning you, however silly or insignificant you think they may seem. If any professional says something you don't understand, ask them to explain what they mean.
- Support groups are a great source of information. If your child has lots of features of a condition, support groups for that condition may be able to help you find information and the right specialists to make a diagnosis.
- If you feel that you need to see a specialist, or if you think you may have somehow got 'lost in the system', do not be afraid to phone. Ask to speak to the consultant's secretary to find out what is happening with your child's appointment.
- Be persistent. If you do not feel all the ways of trying and get a diagnosis for your child have been explored, you can request a second opinion. You can go back to your GP and ask to be referred to a different specialist. Or if you are unhappy with your GP you can ask to see another GP.

National Autistic Society (NAS)

Helpline: 0808 800 4104

Email: nas@nas.org.uk

Web: www.autism.org.uk

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 have been diagnosed with autism, NAS has useful information.

SCOPE

Freephone helpline: 0808 800 3333

Email: helpline@scope.org.uk

Web: www.scope.org.uk

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

Syndromes without a name (SWAN) UK

Tel: 0207 704 3141

Email: SWAN@geneticalliance.org.uk

Web: www.undiagnosed.org.uk

Run by Genetic Alliance UK, SWAN offers support and information to families of children with undiagnosed genetic conditions. They have an online community of families, a blog and a newsletter.



The Communication Trust

Tel: 0207 843 2526

Email: enquiries@thecommunicationtrust.org.uk

Web: www.thecommunicationtrust.org.uk

The Communication Trust is a

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

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Glossary

Additional needs

This term is used to refer to 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.

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

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.

Genetics service

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 11).

Global developmental delay

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.

You can read our guide, *Developmental delay* for more information. Call our freephone helpline on 0808 808 3555 and ask for a free copy.

Metabolic disorder

Metabolism is the process your body uses to make energy from the food you eat. A metabolic disorder occurs 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 others that you need to stay healthy.

Health visitor

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

An occupational therapist pays particular attention to hand-and-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

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

A physiotherapist helps in the management and development of

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movement skills. This may include using exercises to strengthen weak muscles and games to improve coordination and motor skills.

Prematurity

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

The European Union says a condition is 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

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 can call our freephone helpline for information on 0808 808 3555.

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 specialty 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)

Children with special educational needs, (or additional learning needs (ALN) in Wales), have a considerably greater difficulty in learning than others of 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 the person at a school or in early years' settings who has responsibility for coordinating SEN provision.

Speech and language therapist (SALT)

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.

Written by Cheryl Lenny,
February 2013.

Social networking

Contact a Family is on Facebook
and Twitter. Join us at:

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www.facebook.com/contactafamily

Twitter

www.twitter.com/contactafamily

Videos

You can watch videos on our
YouTube channel at:
www.youtube.com/cafamily

Freephone helpline: **0808 808 3555**
www.cafamily.org.uk

Getting in contact with us

Free helpline for parents and families

0808 808 3555

Open Monday to Friday,
9.30am–5pm

Access to over 170 languages

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

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Fax **020 7608 8701**

Email **info@cafamily.org.uk**

Web **www.cafamily.org.uk**

Other guides available

This guide is one in our series 'About diagnosis' for parents and carers concerned with the care of disabled children. Other guides in the series include:

- *Developmental delay*
- *Living with a rare condition*

Contact a Family publications can be downloaded from our website at www.cafamily.org.uk

Parents can call our freephone helpline on 0808 808 3555 and ask for a copy of any of our guides.

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