Living without a diagnosis: for parents of disabled children

Information for families

Incorporating The Lady Hoare Trust

UK
Introduction
This guide is for parents and carers who think their child has a rare condition but do not have a diagnosis. 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, parents and carers can go through a range of emotions. Parents may feel anxious about what the future holds and the tests and assessments your child has can be confusing.

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. Other leaflets in the About Diagnosis series:
  ▪ A genetic condition in the family.
  ▪ Developmental delay
  ▪ Getting a diagnosis
  ▪ Living with a rare condition
  ▪ Support for parents.
Living without a diagnosis

Once you identify that your child may be developing differently from other children you may begin your search for a diagnosis – a reason why your child is the way they are. However, getting a diagnosis might take some time.

The Genetic Alliance UK estimates that about half (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.

The causes of disability

There are many reasons why children are born with a disability, 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 – research has shown that these are more likely to occur when the parents are older or with inter-family marriages
- asphyxia, a lack of oxygen or trauma, during, or shortly after birth.

Parents may worry that it might be their fault that their child has a disability because of something that happened before the baby was conceived or while the mother was pregnant. It is rarely anyone’s fault, though.
# Trying to get a diagnosis

## Why is it sometimes hard to get a diagnosis?
There are a number of reasons why getting a diagnosis can be difficult:

- **there are more than 6,000 known rare conditions.** Doctors rarely see, or read about children with these conditions, making it harder to recognise them when they do
- **different 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**
- **there are significant variations in the way that one condition can affect different children, which might of making a diagnosis**
- **certain things that indicate a condition may not appear until your child is older – this may result in a late diagnosis or even a change in diagnosis**
- **for many conditions the cause is not known.**

## Who can help?
A **health visitor** may be a vital source of support for parents with children under five years. Many parents first discuss their concerns about their child’s development with their health visitor.

For other families, the first point of contact if they have concerns about their child will be their **General practitioner (GP)**. Your GP may refer your child to a **paediatrician** – who specialises in conditions affecting children and young people.

They in turn, may contact a specialist (also called a consultant) – who may look at a particular area of the body or a particular group of conditions. If your child’s symptoms are unusual, then you might find your child is referred to lots of different specialists to see if any of them can make a diagnosis.

Other health professionals may be involved 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 time for parents.
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 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, mainly because of advances in medical knowledge and improvements in testing techniques and medical equipment.

If it is suspected that the cause of your child’s difficulties 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. See our guide in the About Diagnosis series, A genetic condition in the family for more information.

Because of improvements in science and medicine, there may be a belief that doctors can always find out why something has happened to your child. Sometimes this isn’t the case and this can be disappointing.

What to do if you are not happy
If you feel strongly that all ways of getting a diagnosis 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.

Freephone helpline: 0808 808 3555
www.cafamily.org.uk
If after seeing a consultant/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 also explain the reason 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 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 had been at first. For these parents, the most important thing is ensuring that their child’s day-to-day needs were met.

You may, however, want a diagnosis so you can make sure that your child has the necessary follow-up support.

**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 rather than their diagnosis. However, in practice you may find it harder to get this for your child. 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’re entitled to have a social services assessment of your child’s needs and support needs and of your needs as a parent/carer, whether your child has a named diagnosis or not. You can request an assessment by contacting your social services department – in some places there may be a ‘Children with Disabilities team’ who you can contact. Your GP, paediatrician or health visitor may also ask on your behalf.
- You are entitled to receive benefits such as Disability Living Allowance depending on the level of support or care that your child needs. The Contact a Family helpline can offer advice on this.
- Parents also worry that assessments and claims for financial support would not be taken seriously if their child is listed as ‘undiagnosed’. Ask your specialist or paediatrician to write a
letter to outline the features or symptoms your child experiences and what effect this has on daily life to support your claims.

- Your child is entitled to have extra 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.

**How Contact a Family can help you**

Contact a Family can provide support to families even if their child does not have a diagnosis. Services include:

- providing advice, support and information to any parent or carer of a child with additional support needs through our freephone helpline on topics such as benefits, short breaks, aids and equipment and play and leisure, and education
- providing approved medical information on many conditions, including rare conditions. See our website for medical information online at www.cafamily.org.uk/medical-information/conditions
- putting parents in touch with condition support groups, including SWAN UK a support group for families with children who don’t have a diagnosis)
- putting parents in touch with local support groups and other families through their online family linking scheme – MakingContact.org at www.makingcontact.org
- offering a one-to-one family linking scheme for rare conditions, where there is no UK support group
- supporting groups of parents and carers to get together and form their own support group.

**Communicating your child’s needs**

Repeatedly having to explain your child’s medical problems to different doctors can be frustrating. You can make a list of all the way you child is affected and take copies along to all medical appointments and assessments. You can also keep a

“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.”

Mother

Freephone helpline: 0808 808 3555
www.cafamily.org.uk
record of your child’s progress using the Early Support Development Journal.

If your child has complex needs, then explaining these every time you go into hospital or have an appointment can be frustrating. You can create a communication passport for your child to pass on crucial information about a child like information about their additional needs, medications, likes and dislikes and information should an emergency situation occur. The Foundation for People with Learning Disabilities and Bristol Children’s Hospital both produce good templates for hospital passports.

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 features 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 by the child’s specialist or paediatrician to list the child’s needs and how best they can be supported.

“I’m tired of having to explain to people that my child isn’t naughty but has an unnamed condition.” Kate, mother of Jake, 6

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 (SEN) is the early years or school’s special educational needs coordinator (SENCO). To find out more about SEN call Contact a Family’s freephone helpline on 0808 808 3555.

Meeting other families
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 England have a local parents’ support group where families of children with all kinds of disabilities meet up. Local groups have the advantage of meeting regularly and locally, which can provide a support network if you
need one. You can ring Contact a Family’s freephone helpline for local group contact details.

**National condition support groups**

*Syndromes without a name (SWAN) UK* is a national network for families of children with undiagnosed genetic conditions, which is run by the Genetic Alliance UK.

It is worth remembering that whilst your child may not have a named diagnosis, condition support groups may have information that might be useful to you. For example, the *National Autistic Society* may be able to offer useful resources for helping with behavioural problems, *ICAN* and *The Communication Trust* will have information on speech and language techniques and *SCOPE* can help provide solutions to issues with poor muscle control.

**Useful organisations**

*Bristol Children’s Hospital*

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’

Tel: 0117 923 0000

*Early Support*

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 booklet on behaviour and sleep. They also produce the Early Support Development Journal.

Tel: 0207 843 6350
Email: earlysupport@ncb.org.uk
Web: www.ncb.org.uk/early-support

*Foundation for People with Learning Disabilities*

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.

Web: www.learningdisabilities.org.uk

(continues on page 11)
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 the ways your child is affected by their disability or condition. 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 can 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 the specialist about your child. Lots of hospitals now have hospital or communication passport templates.
- Get a diary or a big wall calendar with loads of space to mark appointment times.
- Keep a paper trail. 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 the questions you want to ask at an appointment beforehand. Write these all down in an appointment notebook and take them along with you.
- 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 especially if you are upset.
- 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 and support. If it is suspected your child has a particular condition or you feel that your child has lots of feature of a condition they may be able to help suggest ways of getting 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 doctor’s secretary to find out what is happening with your child’s appointment.
- Be persistent, if you do not feel all the ways to try and get a diagnosis for your child have been explored then 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.
**Genetic Alliance UK**
Genetic Alliance UK is an umbrella body that represents many individual 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.

Tel: 0207 704 3141  
Email: contactus@geneticalliance.org.uk  
Web: www.geneticalliance.org.uk

**ICAN**
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.

Information line: 0845 225 4073  
Email: info@ican.org.uk  
Web: www.ican.org.uk

**National Autistic Society**
National Autistic Society (NAS) is the leading UK charity for people with autism (including Asperger's syndrome) and their families. They provide information, support and pioneering services, and campaign for a better world for people with autism. Even though your child may not be autistic, NAS has information that is very useful for parents.

Autism helpline: Call 0808 800 4104  
Email: nas@nas.org.uk  
Web: www.autism.org.uk

**SCOPE**
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.

Freephone helpline: 0808 800 3333  
Email: response@scope.org.uk  
Web: www.scope.org.uk

**Syndromes without a name (SWAN) UK**
This project, run by the Genetic Alliance UK, offers support and information to families of children with undiagnosed genetic conditions. They have an online community of families, a blog and a newsletter.

Tel: 0207 704 3141  
Email: SWAN@geneticalliance.org.uk  
Web: www.geneticalliance.org.uk/projects/swan
The Communication Trust
The Communication Trust is a campaigning voice for children with speech, language and communication needs. The Trust has a number of useful resources and runs Talking Point and Talking Trouble, which are websites with information for parents.

Tel: 0207 843 2526  
Email: enquiries@thecommunicationtrust.org.uk  
Web: www.thecommunicationtrust.org.uk

Glossary

Additional needs
This term is used to refer to any additional support beyond services available to all children of the same age 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 for an emergency situation.

Disability Living Allowance
Disability Living Allowance (DLA) is the main benefit for children and adults with a condition or impairment. It helps to meet the extra costs that you might have as a result of your child’s impairments. It is made up of two parts, a care component and a mobility component.

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 often caused by new changes to DNA and chromosomes.

Genetics service
There are 23 NHS genetics centres in the UK. These 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 get 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.

**Metabolic disorder**
Metabolism is the process your body uses to get or 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 other ones 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 involvement with children with disabilities.

**Occupational therapist**
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.

**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.

**Physiotherapists**
A physiotherapist helps in the management and development of movement skills. This may include exercises to strengthen weak muscles and games to improve coordination and motor skills.
Prematurity
The definition of 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. There are different levels of prematurity and these carry their own risks.

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
A social services assessment is to gather information about a child or family so they can decide what help they need. Usually an initial assessment will take place with a more detailed assessment taking place later. The process for assessment is in the Assessment Framework, which is statutory guidance that the local authority must follow.

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 neuroloist for conditions affecting the brain. Once specialty training has been completed, doctors are able to apply to consultant posts. Consultants accept ultimate responsibility for the care of patients referred to them, so it is a position of considerable responsibility.

Special educational needs
Children with special educational needs (SEN) 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.

Specialist 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 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:

Facebook
www.facebook.com/contactafamily

Twitter
www.twitter.com/contactafamily

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

Getting in contact

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:

- A genetic condition in the family
- Developmental delay
- Getting a diagnosis
- 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.