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This Factsheet is one of a series produced for parents and groups concerned with the care of disabled children. A full list of Contact a Family publications is available on request.

incorporating  
**The Lady Hoare Trust**



## Introduction

This factsheet is aimed at parents who are caring for a child who does not currently have a diagnosis. It may help to answer some of the questions you may have; more importantly it will highlight that you are not alone. There are many other families in a similar situation, all searching for information that will help them understand their child's particular symptoms and needs and ensure that they receive the help, support and services they need.

*"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 really matter. All the professionals involved with Helen only needed to know what her immediate difficulties were so that the right care could be arranged."*

When you discover your child has a disability, your emotions can be very varied and intense. Parents commonly describe feelings of isolation, anxiety, sadness, despair, guilt and anger mixed up with an overwhelming love and protectiveness.

It is natural to want to know the cause of your child's disability. Without a clear diagnosis you can feel in limbo, trying to come to terms with what is happening and unsure what the future holds for your child and the rest of the family. Many parents describe their feelings of frustration as they are referred from one specialist to another, desperately seeking a diagnosis. Finding ways around the system to access information, help and support can be confusing and parents often worry that their child will be deprived of appropriate services because they have no "label".

There are no easy solutions and although some families receive a diagnosis at a later stage, for others, sadly, this does not happen.

## Why did it happen?

There are many reasons why children are born with disabilities and/ or health needs. These include:

- ☞ Difficulties during the pregnancy including infections in the mother;
- ☞ prematurity;
- ☞ environmental problems;
- ☞ genetic conditions;
- ☞ asphyxia or trauma during or shortly after the birth.

Although in many cases it is almost impossible for paediatricians to single out a specific cause, children will be left with a number of problems that do not fit into any one condition. Sometimes parents who have a child with an apparently obvious condition can discover that the cause is quite different from what they believed it to be.

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



### The problem with statistics

It has been estimated that there are 696,000 disabled children under the age of nineteen years<sup>1</sup>. However it is difficult to know how many of these children do not have a diagnosis. Problems with identifying and diagnosing very rare conditions, misdiagnosis and late diagnosis have added to the difficulties of knowing how many children come into this category. Attempts have been made to estimate numbers but lack of research has made it impossible to be accurate about figures.

## Why are some conditions so hard to diagnose?

There are a number of reasons why making a diagnosis is not easy:

- Doctors are now seeing a large number of children with very rare conditions, which are sometimes difficult to identify;
- Many conditions have similar features and symptoms, which make it difficult to be accurate about a diagnosis;
- Some children will have a number of problems that do not fit into one specific condition;
- There are substantial variations in the degree to which a child may be affected by a disorder or syndrome, which might add to the problem of making a diagnosis;
- Certain identifying features may not appear until later in the child's development resulting in a late diagnosis or even a change of diagnosis;
- Identifying a medical professional who specialises in particular conditions can take time.

Another factor is the multitude of new syndromes that are being identified. Parents who may once have been told simply that their child was "developmentally delayed" might now have different syndromes suggested as possibilities. This can raise hopes of a precise diagnosis – if only you could find the right expert.

## Is it genetic?

Genetics is a branch of biology concerned with heredity and individual characteristics. Some specific conditions and rare syndromes have a genetic basis. There are a variety of reasons why genetic conditions arise including structural or numerical changes in chromosomes, changes (or mutations) in a single gene or changes (or mutations) in a number of genes. Some conditions tend to run in families whilst others may occur for the first time in a family (sporadic).

The field of clinical genetics is rapidly advancing and it is now possible to diagnose a large number of conditions using a variety of testing methods available. If there is evidence of a specific condition amongst family members, the option of genetic testing may be discussed. This may help to confirm or rule out whether your child's difficulties are the result of a genetic condition. Your GP or paediatrician are in a position to refer you to your nearest Regional Genetics Centre or you can self-refer.

Contact a Family produce "A genetic condition in the family", a factsheet which gives a brief introduction to genetics, explores what genetic counseling involves and tries to address many of the worries and concerns that surround it.





## Living without a diagnosis

*“A sense of helplessness and frustration may be eased if parents can be encouraged to keep a diary as an ongoing picture of their child’s development. This may not alter how things are, but it may give a perspective to health professionals and others who monitor the child. Parents may also need to record their own thoughts and feelings.”<sup>2</sup>*

A diagnosis can give a good indication of how a child may or may not develop. Without this information parents fear that it will be difficult to obtain suitable services. It might be helpful to bear the following points in mind:

- ☞ In any assessment, focus on the child’s current symptoms.
- ☞ It may help to have a supporting letter from your child’s paediatrician confirming the current symptoms.
- ☞ Consider keeping a diary of appointments, extra help and attention needed by the child and all the demands on your time and finances. Record details of the child’s milestones and current stage of physical and mental development in relation to able-bodied children.
- ☞ It helps if an annual review can be provided in the light of the child’s developing condition. Use any such reviews as an opportunity to express your views of how your child is developing and his or her current needs, and to re-think any earlier decisions if necessary.
- ☞ It is helpful if you can avoid confrontation with service providers and find ways of working in co-operation with them. At the same time you may have to be persistent – parents often say that they received help only because they have been prepared to speak up and battle for services.
- ☞ Under various pieces of legislation throughout the UK, you are entitled to ask for an assessment of your needs as a carer and your child’s needs.

## Exploring all avenues

Some families feel it is important to explore all avenues when seeking a diagnosis for their child. One of the greatest difficulties for parents in this situation is identifying the medical professional who may be able to advise them on the options available. Some children will be under the care of the local hospital whilst others will be seen at a specialist centre. Children with a number of differing problems may be under the care of more than one professional worker. Dealing with the medical professionals can be frustrating due to limited appointment time, infrequent outpatient appointments, lack of information and not always having access to a consultant. It pays to be prepared!

- ☞ Make a list of the questions you want to ask;
- ☞ Request a longer appointment time;
- ☞ Take a friend or family member with you so that they can take notes;
- ☞ Ask for any written information that may help you to understand your child’s specific problems;
- ☞ If possible request an appointment with the consultant without your child being present so that you can have a discussion without being distracted;
- ☞ If your child is under the care of the local paediatrician do ask if there are any other options available to you. This could include a referral to a specialist centre or specialist paediatrician but you may have to travel long distances for appointments.

If you feel strongly that all avenues to getting a diagnosis have not been explored, do discuss this with your child’s paediatrician. Parents have no legal right to a second opinion but they can request one. It is not, however, guaranteed to provide you with further information nor a firm diagnosis. Families in England can ask their local PALS (Patient Advocacy and Liaison Service) for more advice about this or any other health issue. Community health councils in Wales, health and social services councils in Northern Ireland and health councils in Scotland provide similar support.

For further information about the assessment process Contact a Family produces ‘Assessments and Services in England and Wales’ and ‘Assessments and Services in Scotland’. Contact our Helpline (0808 808 3555) for a copy.

## Influencing the services your child receives

You may find that arranging services for your child without medical diagnosis raises issues which parents of children with a diagnosis do not come across. There is an increasing shift in government policy and legislation towards parents of disabled children being able to be involved and influence how services are delivered. If you would like to get involved in influencing your local service providers and make them more aware of the needs of you and your child, make contact with other parents of disabled children in your area to see if you can work together. For more information about parent participation in service provision please get in touch with the Contact a Family helpline.

## My unique child

Some parents, after several years, do achieve a diagnosis for their child only to find that the label does not matter anymore. With or without a diagnosis, all parents of disabled children face the challenges in accessing adequate services, and the same heartaches and joys of having a special child.

For many parents the best support comes from other parents in local multi-disability groups that typically represent a range of physical and learning disabilities, some with labels and some without. Contact a Family can provide information about joining or starting a local group. Call the Contact a Family local groups officer, Tel: 020 7608 8700 for more information. SWAN (Syndromes Without A Name), the UK support group (see page 7) can also provide information and support.

In any gathering of parents of children with mixed disabilities it can be guaranteed that a few will say:

“I did not get a diagnosis until Amy was 12” or “I have never had a diagnosis for Sammy”. However, they will all talk of the same needs for information and support and invariably add that their greatest help has come from other parents of disabled children.

*“When the most recent diagnosis tests came back normal, I decided that it was time to accept that Chrissie is unique. I have support from her school and from other carers who give us respite. We have to deal with each new issue in the best way we can. Now I am not so fixated on the idea of a diagnosis as a solution, I can get on with my own life. When Chrissie goes through one of her troublesome phases I know it will not last forever.”<sup>3</sup>*



## Investigations

There are a number of tests and investigations that can be used to help determine the way in which your child is affected. These tests may help in obtaining a diagnosis. The most common tests and investigations are:

**Blood Tests** – These can be used to help identify many things such as the number of red and white blood cells, infections, the amount of oxygen in the bloodstream and the child’s blood group. Chemical imbalances indicating a possible metabolic disorder may be identified by blood tests.

**Chromosome Studies** – These can give detailed information of the chromosome structure. They are only available in specialist centres.

**Computerised Tomography (CT) Scan** – This scan gives detailed images of the inside of the skull and the body.

**Developmental Tests** – During early childhood all children have developmental tests. These start from birth and are then followed by regular routine assessments of height, weight, head circumference, reflexes, co-ordination, speech, hearing, sight and physical development. These can be the first indication that a child is not reaching the expected milestones.

**Electroencephalogram (EEG)** – Records the electrical activity in the brain.

**Gene tests** – These may give detailed information about changes (or mutations) in the gene or genes. Such tests are usually carried out at regional genetics centres.

**Magnetic Resonance Imaging (MRI) Scan** – Gives a detailed picture of the internal organs of the body using a computer.

**Ultrasound** – Produces a detailed image of organs such as the kidneys, abdomen and liver onto a screen, which is then recorded on film.

**X-rays** – Used to give a detailed image of the bone structure of the body.

## Who's who

For parents one of the most confusing areas is that of understanding the roles of the various professionals involved in the care of their child. These are some you may have contact with:

**Audiologist** – Works with children who have hearing difficulties and can advise on aids to improve hearing.

**Care Manager** – Assesses, organises and reviews the total care required for an individual. Can work for the local Social Services or the Health Authority. Please note that this is a generic term and that decisions are made by more than one individual.

**Children's Community Nurse** – Not available in all areas. Responsible for carrying out certain nursing procedures and treatments at home. Can also advise families on caring for their child at home.

**Clinical Psychologist** – Works with the child and family to help with any behavioural and developmental problems.  
**Community Paediatrician** – Co-ordinates community health services and makes relevant referrals to other agencies. May be involved with educational assessments.

**Consultant** – The most senior member heading a team of doctors. In overall charge of a child's care.

**Continence Adviser** – Provides information, advice and support to promote continence and the management of incontinence. May also supply continence aids. Some areas have Enuresis Clinics to help with daytime and night-time wetting.

**Dietician/ Nutritionist** – Will offer advice and help on special diets and feeding.

**Educational Psychologist** – Works with parents and teachers in assessing the psychological and educational needs of children with any learning difficulties.

**Geneticists** – Doctors and genetic counsellors (who may have a nursing background). Provide information about the likelihood of a genetic condition happening in a family and about the medical management of a condition.

**Key Worker** – Maintains regular contact with your family. Ensures that information about your child is shared efficiently with everyone who is working with your family. However, they are not yet available in all areas.

**Occupational Therapist** – Will help and advise on special aids and equipment, which will help in all areas of mobility and daily living.

**Orthoptist** – Works with children who have vision problems, abnormal eye movements and can check for squints.

**Paediatrician** – Doctor who specialises in the care and treatment of children whilst in hospital or Out-Patients Department.

**Physiotherapist** – Using exercise and movement will help the child to gain as much independence as possible.

**Social Worker** – Provides help and advice as well as emotional support with social and financial problems. Will also help to obtain local services such as respite care.

**Speech and Language Therapist** – Works with children who are experiencing speech or language problems to enable them to communicate. Also helps with other difficulties such as feeding caused by oral problems.

## Specialities

Hospitals will vary in the range of conditions they are able to treat. Below is a list of some of the specialist departments and the functions they cover:

**Cardiology** - heart and circulation;

**Dermatology** - skin;

**ENT**- ears, nose and throat;

**Endocrinology** - any internal organs which produce secretions, such as the thyroid gland, stomach and liver;

**Gastroenterology** – oesophagus, stomach and intestines;

**Haematology** – blood;

**Immunology** – the body's reaction to dealing with bacteria and viruses;

**Nephrology** – kidneys and the urinary tract;

**Neurology** – brain and nervous system;

**Ophthalmology** – eyes;

**Orthopaedics** – bones and joints;

**Psychology** – study of behaviours;

**Respiratory** – lungs and associated organs which involve breathing;

**Rheumatology** – joints and muscles.

## References and Acknowledgements

[1] Family Resources Survey (FRS), (2002/2003)

[2] Helen Vegoda, Counsellor in Child & Family Support, Bristol Royal Hospital for Sick Children

[3] The last tests came back normal, Jane Gregory, Disability Now, February 1997.

And the parents who kindly allowed us to quote their experiences.



## Support groups

### SWAN (Syndromes Without A Name)

6 Acorn Close

Great Wyrley

Walsall

WS6 6HP

Tel. (01922) 701234

e-mail: [info@undiagnosed.org.uk](mailto:info@undiagnosed.org.uk)

Web Site: <http://www.undiagnosed.org.uk>

*This group offers a 24-hour helpline with a listening ear and, wherever possible, links with other families. It publishes a newsletter three times a year, information on possible therapies and a reading list. It also offers an information pack covering issues such as benefits, mobility and questions to ask a geneticist. The group has international links.*

## Other Support

Children's Centres may be able to offer you support. Sure Start is a government programme which aims to achieve better outcomes for children, parents and communities. Sure Start Children's Centres are places where children under five years old, and their families, can receive integrated services and information. To find a Children's Centre in your area you can search the Children's Centre Online database at:

Web: <http://www.tinyurl.co.uk/cvsi>

The Department for Education and Skills (DfES) has produced the Early Support Family Pack. As a part of this pack, one of a series of booklets '*When your child has no Diagnosis*' can be downloaded from the following website or ordered online:

Web: <http://tinyurl.co.uk/zl2r>

If you do not have internet access please get in touch with our Helpline (0808 808 3555) as we can post you the information.

A leaflet entitled '*Finding Medical Information on the Internet*' has been produced by Contact a Family and approved by the Royal College of Paediatrics and Child Health. For a copy please visit our website ([www.cafamily.org.uk](http://www.cafamily.org.uk)) or phone our Helpline (0808 808 3555).

### MakingContact.org

Parents with children who do not have a diagnosis can make a request on <http://www.makingcontact.org> to find others with similar undiagnosed conditions e.g. undiagnosed bone conditions or undiagnosed metabolic disease.

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