

# Undiagnosed Children fact sheet

This fact sheet 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 special needs and ensure that they receive the help, support and services they need.

When you discover your child has special needs or 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.

## The problem with statistics

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 did it happen?

There are many reasons why children are born with special 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 pediatricians 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 the cause is quite different from what they believed it to be.

# 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 that specializes 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 "mentally handicapped" 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, you may be offered genetic testing for your child. This may help to confirm or rule out whether your child's difficulties are the result of a genetic condition. Your GP or pediatrician will refer you to your nearest Genetics Center.

## 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 and seen at different specialist center. 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 pediatrician do ask if there are any other options available to you. This could include a referral to a specialist center or specialist pediatrician but you may have to travel long distances to other centers.

If you feel strongly that all avenues to getting a diagnosis have not been explored, do discuss this with your child's pediatrician. You can ask for a second opinion or change pediatricians however; it is not guaranteed to provide you with further information nor a firm diagnosis.

## Living without a diagnosis

Common things that are said or thoughts from those who are dealing with the frustrations of not having a diagnosis.

**-Will future children be at risk?** This is the most common concern of parents with undiagnosed children. The amount of risk you are taking in having another child, could be the deciding factor. Or preparing yourself of the risk is very important to those wanting more children. If your child is able to have children will their child be effected? Or if siblings have children will their children be at risk.

**- It is difficult to access support services.** Reasons may be not knowing where to turn. Finding support in just one area of your child's differences may leave parents feeling alone because their child has other differences that the supporters can't relate.

**- Unable to get any information about the child's disorder.** Learning everything there is to know about something that affects your child is very important to many. When unable to do this it may leave some feeling like they aren't doing everything that they can to support their child.

**- Always debating whether to test or not.** Not knowing what might occur leaves the doctors either testing too much when it is not necessary or not enough when it is important. Many times this leaves parents in a very hard and emotional position. When tests keep coming back negative, parents may start to refuse some test that may be helpful. Having to put children through all the negative testing is many times very traumatic for both child and parent. At least if the test was positive what you put your child through might feel a little easier when knowing the possibility of a positive outcome that might come from knowing.

**- Parents are looked at as the problem.** In some cases Parent are thought to be the cause of the child's problems. This adds to stress that some parents are already dealing with. This could cause more difficulty in dealing with the real issues that the child may be having.

## 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 children

with disabilities face the same hurdles of battling for 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, or SWAN (Syndromes Without A Name). 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 children with special needs.

## **Who's Who?**

For parents one of the most confusing areas is understanding the roles of the various medical professionals involved in the care of their child.

These are some you may have contact with:

- Educational Psychologist works with parents and teachers in assessing the psychological and educational needs of children with any learning difficulties.
- Occupational Therapist will help and advise on special aids and equipment, which will help in all areas of mobility and daily living
- Physiotherapist use exercise and movement will help the child to gain as much independence as possible.
- Dietician/Nutritionist will offer advice and help on special diets and feeding.
- Speech and Language Therapist Works with children who are experiencing speech or language problems to enable them to communicate.
- Audiologist works with children who have hearing difficulties and can advise on aids to improve hearing.
- Social Worker Provides help and advice as well as emotional support with social issues.

## **Specialties**

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:

- Anesthesiology - sedation
- Cardiology - heart and circulation
- Dermatology – skin
- Endocrinology - any internal organs which produce secretions, such as the thyroid gland, stomach and liver
- ENT- ears, nose and throat
- Gastroenterology - esophagus, stomach and intestines
- Genetics - look at all features as a whole, to determine a possible cause
- Hematology - 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
- Orthopedics - bones and joints
- Psychology - study of behaviors
- Respiratory - lungs and associated organs which involve breathing
- Rheumatology - joints and muscles

# Investigations

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

- Developmental Tests- During early childhood many children will have a developmental test. 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.
- 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.
- Chromosome Studies- These can give detailed information of the chromosome structure. There is an overall analyzes that gives a big picture, then there is a FISH probes that look at certain areas of certain chromosomes. There are many research studies checking out many different areas of chromosomes, many times your geneticist will suggest some research studies.
- Computerized Tomography (CT) Scan- This scan gives detailed images of the inside of the skull and the body.
- Magnetic Resonance Imaging (MRI) Scan- Gives a detailed picture of the internal organs of the body using a computer.
- X-rays- Used to give a detailed image of the bone structure of the body
- Ultrasound produces a detailed image of organs such as the kidneys, abdomen and liver onto a screen, which is then recorded on film.
- Electroencephalogram (EEG)- Records the electrical activity in the brain.

## Support Group

SWAN USA (Syndromes Without A Name USA)  
Amy Clugston  
1745 Lorna Lane  
Otsego, MI 49078  
amyclugston@undiagnosed-usa.org  
Web Site: <http://www.undiagnosed-usa.org>

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