contact a family
for families with disabled children

About diagnosis: Getting a diagnosis

Understanding the causes of disability and the process of diagnosis

- Blood tests
- Genetic counseling
- Hearing assessments
- Scans
- Ultrasounds

Part 5

30 NOT OUT

30 years of strengthening families with disabled children

Although some children may be diagnosed before or very soon after birth, many children's condition may not be noticed straight away. As a parent you might be the first to notice that your child seems not to be developing quickly enough or is developing differently from other children.

Medical or health professionals may feel at this point that it's necessary to monitor a child's development. Initial tests will be carried out which could include hearing and sight tests, or other assessments of a child's development. If a particular genetic disorder is suspected by a health professional (see section 'What is a genetic condition?) there may be a referral for specific tests. The exact tests or investigations will depend on what the underlying cause of disability is known or suspected to be.

Causes of disability

Accident or trauma
Your child may have been in an accident which has caused them to damage or even lose a limb which can cause additional needs.

Difficulties during pregnancy or birth
Complications can sometimes occur during both pregnancy and labour. These may result in temporary or permanent problems.

Babies born prematurely
When babies are born early their developing brains can be easily damaged, and they are also at increased risk of infection and injury. Sometimes blood flow and oxygen to the brain are restricted which can cause problems with vision, movement and development.

■ **Environmental problems**
A disability or health condition can be described as having environmental causes when an unborn child is affected by a mother's exposure to radiation or chemicals, or if the baby is affected by a mother's health problem in pregnancy.

■ **Genetic conditions**
For our bodies to work we need many thousands of genes to work together. Changes in different genes can result in many different conditions or disabilities. See section 'What is a genetic condition?' for more information.

■ **Unknown**
The exact causes of many conditions are unclear. Some arise from a combination of genetic and environmental factors. This can make it difficult to pin down the exact cause.

Tests and 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, past infections, the amount of oxygen in the bloodstream and the child's blood group. Chemical imbalances indicating a possible metabolic disorder may be identified using blood tests.

■ **Brain imaging** – a Magnetic Resonance Imaging (MRI) Scan gives a detailed picture of the brain and internal organs to look for abnormalities. A Computerised Tomography (CT) Scan can also be used but this is less detailed and best for looking at trauma or internal bleeding.

■ **Chromosome studies** – these can give detailed information of the chromosome structure to identify any chromosome abnormalities that can cause a condition or disability. They are only available in specialist centres.

■ **Developmental tests** – these tests may be done if there is concern about a child's development. They can include assessments of height, weight, head circumference, reflexes, co-ordination, speech, hearing, sight and physical development.

■ **Electroencephalogram (EEG)** – records the electrical activity in the brain. Conditions such epilepsy can affect the electrical activity of the brain and an EEG can be used to identify this.

■ **Electromyography (EMG)** – this test measures the electrical activity of muscles and nerves. It can help to diagnose nerve muscle or spinal problems.

■ **Facial scanning** – computers are used to scan and create a three dimensional image of a child's face to spot subtle variations which can help diagnose a rare disease.

■ **Hearing assessments** – all babies have their hearing tested soon after birth and again at school entry. Other hearing assessments can also be used to gain an indication of hearing loss or problems with development of the hearing system.

■ **Gene (DNA) tests** – these may give detailed information about changes (or mutations) in the gene or genes which can cause or be a factor in a genetic condition. Such tests are usually carried out at regional genetics centres. Blood, skin or cells can be taken for the tests. This will sometimes be done by pricking the heel to get blood or swabbing the inside of the cheek to get cells.

■ **Genetic counseling** – this is a service that provides information and support to families who feel their child or other family members may be affected by a genetic condition. For more information see section 'What is a genetic condition?'.

■ **Occupational therapist assessments** – this is often a series of tests to assess if a child has difficulties with practical and social skills. The tests are designed to identify those things which are a challenge for the child and to find ways to promote improved function in all aspects of life.

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

■ **Vision assessments** – can be carried out as part of the process of diagnosis to determine the amount and type of visual loss or problems. Information gathered through this type of testing can be used to confirm a diagnosis or to assess the needs a child may have. This can help determine any assistance or adjustments, including those to educational provision, that may be required.

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

If you're unsure about the purpose of any treatment or test, ask the person who referred your child or one of the people doing the test.

Other guides in this series

1. How we can help
2. Support for parents
3. What is developmental delay?
4. Understanding behaviour
5. Getting a diagnosis
6. What if we don't get a diagnosis?
7. What is a genetic condition?
8. Rare disorders
9. Sources of medical information
10. Concerned about your child
11. Practical and financial help

Getting in contact with us

Free helpline: **0808 808 3555**
Open Mon–Fri, 10am–4pm;
Mon, 5.30–7.30pm

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

Contact a Family Head Office:
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Tel 020 7608 8700
Fax 020 7608 8701
Textphone 0808 808 3556
Email info@cafamily.org.uk

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Registered in England and Wales No. 1633333
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The Royal College of Paediatrics and Child Health

The Rayne Foundation

Other information available

- NHS and caring for a disabled child (England & Wales)
- Disabled children's services (England & Wales/Scotland)
- A guide to claiming DLA for children (UK)
- Fathers (UK)
- Siblings (UK)
- Grandparents (UK)
- Relationships and caring for a disabled child (UK)

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