

Department of Medical Genetics

Parents' guide to the investigation of children with developmental delay

You have been given this leaflet to give you more information about what might happen during the assessment of your child's developmental delay.

What is developmental delay?

Developmental delay is the term used when a child does not progress as expected. For example, your child's speech may lag behind that of other children or your child may have been slow to learn to walk. In some children, developmental delay is suspected soon after birth because of feeding difficulties or unusual muscle tone. In other children, developmental delay is only suspected much later when learning or behavioural difficulties surface at school.

What causes developmental delay?

There are many different causes for developmental delay. It is a common condition affecting 1 to 3% of the population. Delay may be caused by a child's genetic makeup (for example: Down's syndrome); by problems during pregnancy (such as infection); by problems around the time of delivery, especially if very premature (for example: bleeding in the brain); by problems in early infancy (for example: meningitis) or later in childhood (for example: head injury).

A cause can be found in about half of cases.

Why can it be useful to find a cause?

There are a number of benefits for you and your child to finding out what might be causing the developmental delay. These include:

- Helping you to find the best way to care for your child.
- Helping you, as parents, make contact with a support group and other families.
- Helping your child's health professionals to understand the condition and how it may affect your child in the future.
- Helping your child's health and education and social care professionals to find the best ways to care for your child.

- Addressing concerns you might have about events that happened before your child was born.
- Helping you find out whether any future children you or your family have could also have the condition.

Assessment of developmental delay

A paediatrician (a doctor specialising in children's health) usually does the assessment. The paediatrician will ask questions about your child's medical history, examine your child, measure your child's height, weight and head circumference and assess your child's developmental progress. Other professionals may be involved in this assessment and your child may be referred for hearing or vision assessment.

Your child may be offered some tests or further referrals to try to determine the cause of their developmental delay, for example to a neurologist (a doctor who specialises in the brain and nervous system) or a geneticist (a doctor who specialises in a people's genetic makeup). These specialists will explain what is involved in any further investigations and discuss with you the best way of proceeding. You will be asked to formally give consent to any investigations. If you decide against any further investigation now, the option to have the tests or further referrals at a later stage will remain open in the future.

Tests

There are many different types of test available. These include:

- **Maternal infection tests:** these are to check whether a mother had an infection during her pregnancy that affected her child, for example: rubella.
- **Chromosome analysis:** chromosomes are structures in the body that contain genetic information. A chromosomal disorder can occur spontaneously (a one off occurrence) or it can be inherited from a parent. Looking at the chromosomes can help to identify hundreds of disorders that cause developmental delay.
- **Specific genetic tests:** there are thousands of genetic conditions that can cause developmental delay in a child. In order for the paediatrician or geneticist to select the appropriate tests for your child, x-rays or a brain scan may be necessary.

Usually the tests involve taking blood or urine samples from your child. Occasionally, other samples may be necessary and sometimes a second sample is requested. Samples may also be taken from parents to find out whether either parent is a carrier of the genetic disorder.

Not every child with developmental delay will be offered tests. There are many conditions for which there is no genetic test yet and some conditions are easily recognised by an experienced paediatrician or geneticist so that a diagnosis can be made without the need for further tests.

Clinical Genetics referral

Your child may also be referred to a clinical geneticist who will take a medical history and will want to know if there are any medical problems that run in the family. A family tree will probably be drawn based on the information you are able to provide. Your child is likely to be given a detailed physical examination. The facial and physical appearance of your child may give clues to a diagnosis and photographs (kept confidential in the clinical notes) may be taken. This is so that the geneticist does not have to rely on memory after the appointment.

It is important to remember that not all genetic disorders are inherited from parents as they can arise spontaneously in a child without a previous family history.

When will we get the results?

It varies considerably. Although some test results may come back within a few days, chromosome analysis is likely to take a few weeks. For more complicated or rare conditions, the results can take several months. You can ask your paediatrician or geneticist for an estimate of how long it will take to get the results back. Understandably, parents often find waiting for the test results a stressful time and you could find it helpful to talk to your health visitor during this period.

What if a diagnosis is made?

The paediatrician or geneticist will explain what the diagnosis means for you and your child, for example: what the common features of the condition are, whether there is any treatment for the condition, whether the condition can be passed from one generation to the next.

Even though most parents would like to know the cause of their child's developmental delay, the news may still come as a shock. Parents can find it difficult to concentrate on all the information given to them about their child's diagnosis when they first receive the results.

Many parents find it helpful to have someone they trust be with them when they receive the results such as a partner, close friend or health visitor. You may also want to consider whether it is appropriate for your child to be present at the appointment when the results are given.

A specific diagnosis can be emotionally tough on brothers and sisters too and, especially if the condition is inherited, tensions in the wider family can arise. Emotions experienced by parents vary widely but may include relief, shock, numbness, grief and guilt. If you are adversely affected after receiving a diagnosis, then contact your health visitor or GP.

In time, once these initial reactions have worn off, most families believe that it was worth investigating the cause of their child's developmental delay. Parents report a sense of 'being able to move on'.

What if there is no diagnosis?

In spite of investigations, approximately half of children with developmental delay will not receive a specific name for their condition. In those cases where a name is not given, it is common for parents to have a mixture of feelings:

- Relief that their child does not have the condition that was being tested for.
- Disappointment/frustration that a cause has not been found.
- Isolation as they don't know anyone else whose child has the same condition.

Even if there is no diagnosis, the paediatrician or geneticist may be able to say what the most likely cause of your child's developmental delay is for example: an event during pregnancy or a genetic condition. As medical knowledge improves, you may be given the chance to test your child again. It is important to remember that, with or without a diagnosis, services for your child will not be affected.

And finally.....

Remember that developmental delay does not stop a child from having a full and rewarding life. Your child's health and educational professionals aim to help your child have a life as enjoyable and fulfilling as possible.

If you do not understand anything in this leaflet, contact your health visitor. Your health visitor may also be able to put you in contact with a relevant support group.

You may also wish to contact 'Contact a Family', a national charity, who provides information and support links for families with disabled children.

Telephone: 0808 808 3555, website: www.cafamily.org.uk



We are currently working towards a smoke-free site. Smoking is only permitted in the designated smoking areas.

For advice and support in quitting, contact your GP or the free NHS Stop Smoking helpline on 0800 169 0 169

Help with this leaflet:



If you would like this information in another language, large print or audio format, please ask the department to contact Patient Information: 01223 216032 or

patient.information@addenbrookes.nhs.uk



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