

Some inherited conditions do not usually become obvious until well into adult life and are untreatable. Some adults who are "at risk" of developing such an illness choose to have a genetic test so they can know what is in store for them in the future, but many do not. Having a test helps some people, but others find the result too distressing. For this reason, this department never tests healthy children for these conditions. We believe that decisions about testing in these circumstances should only be made in adult life by the person who is "at risk", after a very careful discussion of their own views and feelings.

Sadly, there are other untreatable conditions that begin in childhood, when a healthy child is too young to make a decision about presymptomatic testing. In these circumstances, although a genetic test may not help a child directly, it may allow the child's family to plan ahead and begin to come to terms with their child's future illness. In such a situation, we would ask to see the child's parents in the clinical genetics department before testing is performed, so that we can help them make the decision that seems best for them and help prepare them for the result of a test that may give a deeply distressing result.

#### A CARRIER test

A genetic carrier test can identify a healthy person whose children could be affected with a particular genetic condition. As such a test has no implications for a person until they come to have a family of their own, testing a child to see if he or she is a "carrier" seldom has any immediate benefits for the child. As a result, it is felt increasingly that children should not be "carrier" tested until they are old enough to understand the implications of the test and to make their own decision about testing.

However, there are many views about the matter, and parents and doctors sometimes have different views about carrier testing. This department does not have a rigid policy on carrier testing in childhood. However as a general principle, we believe that healthy individuals who may carry genetic disorders where

the concern is purely for their future children should have a chance to discuss the various "pros and cons" of being tested before they make a decision about the test. Obviously, in the case of a young child, they are not in a position to be able to have that discussion and make that type of careful decision. However, many teenagers can understand about testing and decide for themselves.

#### Help for parents and children

We know that parents care about their children and want the very best for them. We can also understand how worried parents must feel when they know that their child may suffer from a genetic disease, may develop an inherited illness in the future, or may carry a genetic condition that could be passed on to future generations. It can also be very difficult to know how or when to talk to a child about any of these situations.

If any parent would like further information, or would like to discuss their family situation with us, we would be very happy to try and help. A face-to-face discussion is often best, but some queries can be dealt with by telephone.

#### Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

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# The Genetic Testing of Children



An information sheet  
to be given to  
parents and families

## What is a genetic test?

A genetic test is a special kind of test, usually done on a blood sample, which can detect the presence of an altered gene or chromosome pattern.

## What are genes and chromosomes?

Our **genes** are the unique set of instructions inside our bodies that make each of us individual. There are many thousands of different genes, each carrying a different instruction. As well as determining how we look, our genes control the way each cell or building block of the body works.

Genes lie on tiny structures called "**chromosomes**", rather like beads threaded onto a string. Each chromosome contains thousands of genes.

Our personal genetic "make-up" - the pattern of genes and chromosomes - remains the same throughout our lives: from the moment we are conceived right up to the end of our lives.

## Are there different types of genetic tests?

Yes, there are. Broadly speaking, they fall into three main groups:

- Diagnostic tests
- Presymptomatic (or predictive) tests
- Carrier tests

### Diagnostic tests

The purpose of a diagnostic genetic test is to try and discover the cause of a person's medical problem or problems. If the underlying cause can be found, a suitable treatment for the symptoms of the illness may be possible.

### Presymptomatic (predictive) tests

A presymptomatic genetic test is one that is performed on a person who, although currently healthy, is known to be "at risk" of developing a particular inherited illness sometime in the future.

### Carrier tests

Many of us "carry" an altered gene or chromosome pattern that, although harmless to us, can cause problems in our children, who inherit some of our genes and chromosomes. A genetic carrier test is one that can identify a healthy "carrier" whose children could be affected with a particular genetic condition.

## Should a child have a genetic test?

From a "scientific" point of view, as our personal genetic "make-up" remains unchanged throughout our lives, it does not seem to matter when a person has a genetic test, as the result should be the same.

However, we know that many genetic tests are not "straightforward" in the sense that they can have difficult, long-term consequences for the person being tested. This is why many parents, doctors and other health professionals have major reservations about testing children.

When a genetic test on a child is being considered, it is usually helpful for professionals and parents alike to ask themselves three important questions:

- Is the test in the child's best interests?
- Could the test do the child more harm than good?
- Is it possible to wait until the child is old enough so they can decide for him or herself whether or not to have the genetic test?

The answers to the above questions often depend on which type of genetic test is being considered.

## A DIAGNOSTIC genetic test

If the result of a diagnostic test (one which aims to find out the cause of existing problems) shows a child to have a serious genetic illness, it is upsetting for the whole family. Nevertheless, an accurate diagnosis is often in a child's best interests. He or she may be able to be helped by a particular method of treatment. Other unnecessary investigations can be avoided. In other words, a diagnostic test often does a child far more good than harm. As a result, parents and doctors will often need to decide which tests may be helpful on behalf of a child who is not old enough to make a decision of their own.

## A PRESYMPTOMATIC genetic test

Whether or not a presymptomatic test on a healthy child who is "at risk" of developing a particular inherited illness sometime in the future is in that child's interests depends on the illness itself. In such situations, the most important question is "**can anything be done to prevent the child developing the condition?**"

For example, there is a certain type of inherited bowel cancer that doctors can detect early by looking into the bowel with a special tiny camera for small growths called "polyps". Healthy children who are "at risk" of having the altered gene that causes this condition can benefit from a particular genetic blood test. If they do not have the altered gene, then they can be reassured and can avoid the discomfort of having the camera inserted into their bowel. If they do have the altered gene, then they know that the special regular check-ups are needed so that the cancer can be prevented. As these check-ups are usually unnecessary until a child is about 14 years old, it is often possible to involve the child in the decision about genetic testing.