

**Myotonic Dystrophy Support Group**

Tel: 0115 987 0080

Email: [mdsg@tesco.net](mailto:mdsg@tesco.net)Web: [www.myotonicdystrophysupportgroup.co.uk](http://www.myotonicdystrophysupportgroup.co.uk)**SADS UK - Sudden Arrhythmic Death Syndrome**

Tel: 01277 230642

Email: [info@sadsuk.org](mailto:info@sadsuk.org) Web: [www.sadsuk.org](http://www.sadsuk.org)**Contact a Family**

Helpline 0808 808 3555 or Textphone 0808 808 3556

Freephone for parents and families (Mon-Fri, 10am-4pm and  
Mon, 5.30pm-7.30pm)Email: [info@cafamily.org.uk](mailto:info@cafamily.org.uk)Web: [www.cafamily.org.uk](http://www.cafamily.org.uk) [www.makingcontact.org](http://www.makingcontact.org)**Genetic Interest Group**Provides information about specific genetic conditions and  
contact details of support organisations.

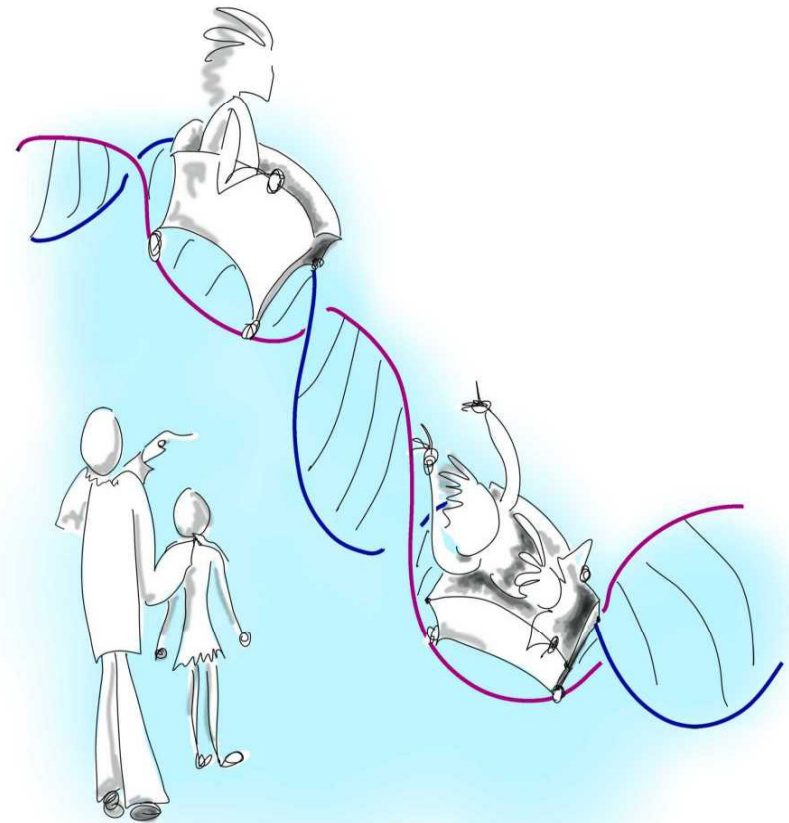
Tel: 020 7704 3141

Email: [mail@gig.org.uk](mailto:mail@gig.org.uk) Web: [www.gig.org.uk](http://www.gig.org.uk)**Genetics and Insurance**More information about genetics and insurance can be found  
online at:[http://www.abi.org.uk/Information/Consumers/  
Health\\_and\\_Protection/499.pdf](http://www.abi.org.uk/Information/Consumers/Health_and_Protection/499.pdf)This information was developed by the Genetic Interest Group, a UK national  
alliance of patient organisations which supports children, families and  
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number 512148.Illustrations by Rebecca J Kent  
[www.rebeccajkent.com](http://www.rebeccajkent.com)  
[rebecca@rebeccajkent.com](mailto:rebecca@rebeccajkent.com)

# Predictive Testing



## Information for Patients and Families

## Predictive Testing

This information is about predictive genetic testing for diseases other than cancer. We have written it to help you answer questions like:

- What is a predictive genetic test?
- Why do some people decide to take one?
- What should I think about if I am considering taking a predictive test?

### Section 1. About our Genes

To understand what a predictive genetic test is, it first helps to understand what genes and chromosomes are.

#### Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 'pairs'. Because the chromosomes are made up of genes, we therefore inherit two copies of most genes - one copy from each parent. This is the reason we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

least 30. In this way you can put the issue aside to look at again in the future.

Once you receive your test results there is no going back. This is why it is important to be very sure about the decision you make, and why it is important to discuss your decision with a trained genetic specialist. Remember that making an appointment with a genetic specialist does not mean that you have to go ahead with testing.

#### Further information

For further information, contact your local regional genetics service. Their contact details can be found at:

[www.gig.org.uk/services.htm](http://www.gig.org.uk/services.htm)

#### Additional information can also be found at:

##### British Heart Foundation

Main telephone number: 020 7935 0185

Heart Information line: 08450 7080 70

Web: [www.bhf.org.uk](http://www.bhf.org.uk)

##### CRY - Cardiac Risk in the Young

Tel: 01737 363 222

Email: [cry@c-r-y.org.uk](mailto:cry@c-r-y.org.uk) Web: [www.c-r-y.org.uk](http://www.c-r-y.org.uk)

##### Familial Spastic Paraplegia

Tel: 01702 218184 Web: [www.hspgroup.org](http://www.hspgroup.org)

##### Huntington's Disease Association

Tel: 0151 298 3298

Email: [info@hda.org.uk](mailto:info@hda.org.uk) Web: [www.hda.org.uk](http://www.hda.org.uk)

##### Muscular Dystrophy Campaign Tel: 0800 652 6352

Email: [info@muscular-dystrophy.org](mailto:info@muscular-dystrophy.org)

Web: [www.muscular-dystrophy.org](http://www.muscular-dystrophy.org)

about yourself and your family when you apply for an insurance policy, particularly above a certain amount. The type of information that insurance companies and employers are allowed to ask varies considerably from one country to another. You should find out whether this includes the results of any genetic tests you may already have taken, or may take in the future. Ask your genetic specialist about it and consult your national legislation.

## Finance

Living with a genetic condition can be difficult financially. Those living with a condition may be unable to work for long periods of time, or may have to stop working altogether. Partners and other family members may also find it difficult to juggle work with the responsibilities of caring for a family member or partner. For some people, knowing that they are at risk of developing a genetic condition allows them time to plan financial and other practical aspects of their future.

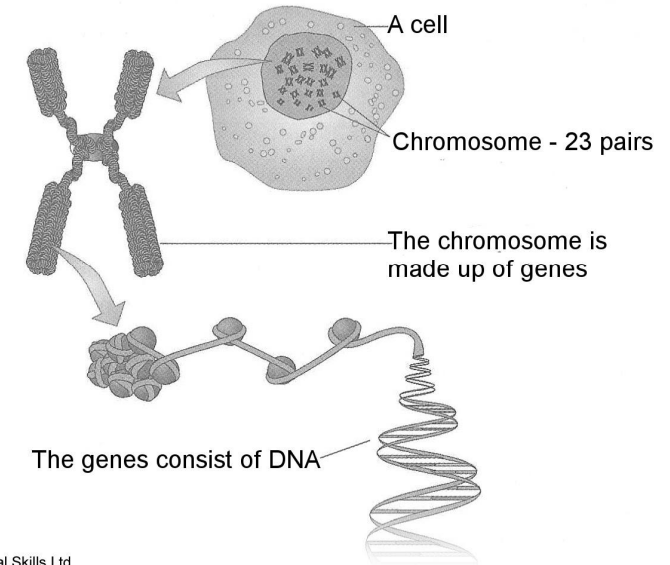


### ***g) Timing of the Test***

If you do decide to go ahead with testing, choose a time when complicating factors from the outside are at a minimum. Divorce, break-ups, stressful times at work, etc. are difficult times to undergo testing, as may be times of celebration such as marriage or childbirth. It is a good idea to plan what you will do the day you receive your results as you may feel emotional, whatever the outcome.

It can be helpful to make a decision about testing even if it is not an absolute, for example, I will definitely not do it until I am at

**Figure 1: Genes, chromosomes and DNA**



Sometimes we inherit or develop a change (mutation) in one (or both) copies of a gene, which stops it from working properly. This change can cause a genetic condition because the gene is not giving the correct instructions to the body. There are thousands of different genetic conditions but some examples are cystic fibrosis, muscular dystrophy and Huntington's disease. Some of these conditions occur from birth. Others occur later on in life. In this leaflet we will mainly be looking at conditions that occur later on in life.

## **Section 2. About the Test**

### **What is a predictive genetic test?**

A predictive test can provide information about whether or not a person will develop or is likely to develop a specific condition, usually at a later stage in life. The test is usually performed on a blood sample. The blood is analysed in a genetics laboratory to

see if there are any changes in the particular gene or genes connected with the disease. In the leaflet ***What Happens in a Genetics Laboratory?*** we provide more information about how genes are analysed.

### **Why might I consider having a predictive test?**

If there is a known genetic condition in your family and the changed gene that causes that condition is known, you might be able to have a predictive test to see if you have inherited the changed gene. You might want to have a test if:

- the condition can be prevented or its symptoms effectively treated, or if
- the condition can neither be prevented, nor its symptoms effectively treated, but:
  - You want the information to help with decisions about having children, or to tell you more about your own children's risks.
  - You believe that knowing more about your chance of getting the disease will help you to make important life decisions including decisions about your health care.
  - You are the type of person that prefers to know more about your own future and prefers to live with certainty than with uncertainty.



how the testing procedure, and the test results, might affect your relationship with your partner and other family members. Remember that an affected family member first needs to agree to be tested so that the changed gene can be identified. Approaching a family member for this reason may be difficult. Sometimes family members have lost touch. It can also be difficult to talk about illnesses that have occurred in the past as it may bring back painful memories. Genetic specialists should be able to offer you advice in these situations.

Some people may want to know about their genetic risk status because of concern for their family members. Other family members, however, may not want to undergo testing because they prefer not to have information about their risk. You will need to be sensitive to this because your test can provide family members with unwanted information about their risk. It is important to remember that members of the same family may have different feelings about testing, and that these feelings should be respected. Genetic testing may sometimes reveal family secrets involving adoption and non-paternity (i.e. the biological father is not who the family thinks it is). This is because the process looks into people's family history and it may become evident that you do not share your genes with your family members. This is a possibility you should be aware of before beginning the process.

### **f) Confidentiality, Insurance and Finances**

#### **Confidentiality**

Access to your genetic test results is confidential. Your doctor is not allowed to tell anyone that you have had a genetic test or to pass on your test results without your permission.

#### **Insurance**

Insurance companies often ask you to provide medical details

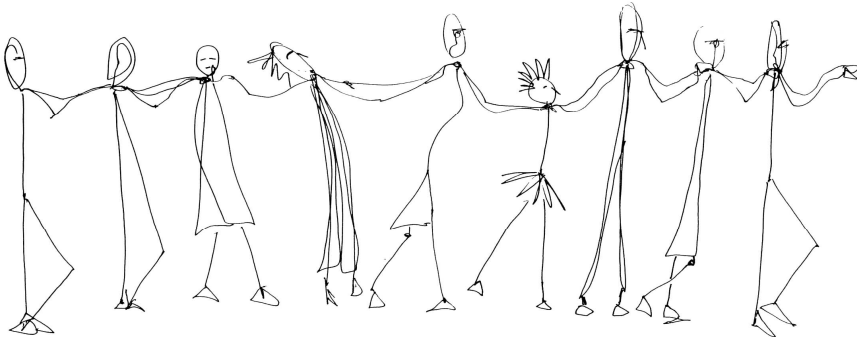
## Is there any way to avoid my child having the condition?

For some genetic conditions, it is possible to perform a test during pregnancy to see if the baby has inherited the changed gene (prenatal testing). For more information you should look at the leaflets about **Amniocentesis** and **CVS**. If you think this might be an option for you, speak to your doctor about whether these tests are available for the condition you are concerned about. It is essential you do this before the pregnancy if possible as the laboratory may have to make preparations that can take several months.

It may be possible to perform a technique called Preimplantation Genetic Diagnosis (PGD) as an alternative to testing the foetus during pregnancy. This involves the couple undergoing medically assisted reproduction, after which the fertilised eggs are tested to see if they have the changed gene. Only those eggs without the changed gene are implanted into the woman's womb. This is a long process and is not suitable for everyone. For more information about PGD, and whether it is available to you, you should speak to your doctor.

### e) Other Family Members

In many cases the genetic testing process brings families closer together and the family can be a good source of support. In some cases, however, the process causes tension and complications within the family. It is a good idea to think through



## Which conditions can be tested for using predictive testing?

There a number of conditions for which predictive testing is currently available. Some examples are:

- Certain types of cancers (for more information please see the **Predictive Testing For Inherited Cancer** leaflet).
- Conditions that affect the nervous system (neurological) including:
  - Huntington disease
  - hereditary ataxia and spastic paraplegia
- Conditions that affect the muscles (neuromuscular) including:
  - myotonic dystrophy
  - facio-scapulo-humeral muscular dystrophy.
- Conditions that affect the heart (cardiac) including:
  - hypertrophic cardiomyopathy (HCM)
  - long QT syndrome

### What do we mean by 'risk'?

In most cases (apart from cancer), having the changed gene usually shows that you will go on to develop the condition. However, it is not usually possible, at least at present, to predict at what age you will develop the disease, how you will be affected, or how rapidly the symptoms will progress.

If you feel you may be at risk and want to consider having a predictive test for one of these conditions, you should make an appointment to see a genetic specialist.

### Genetic testing procedure

Before you can proceed with predictive testing, it must be

confirmed that you are at risk. First, a detailed family history will be done to see if you are at risk of the genetic condition. In most cases, if genetic testing is possible, a close relative of yours who is affected by the condition will be offered genetic testing first in order to identify the changed gene that runs in the family. If a specific genetic change is found in their blood sample then it is possible to take a blood sample from you to see if you have also inherited the same genetic change. If the result of this test shows that you have inherited the same genetic change then you would also be at risk of developing the condition during your lifetime. If you have not inherited the change then you would not be at increased risk. For some conditions such as Huntington's disease, it will not be necessary to test an affected family member first because scientists already know the changed gene to look for.

Taking a genetic test is your choice and you should never feel pressured into taking it by health professionals, family or friends. It is also a long process, which can involve several sessions with the genetic health professional and months waiting for the results from the laboratory. You may be given information that is very new to you and quite complicated, and it can be difficult to take it all in. It's a good idea to take a support person, such as a friend or partner, with you to your appointments. You may want to ask them to take notes during the appointments. It is important that you have the opportunity to discuss genetic testing with a trained genetic health professional. They will be able to provide you with all the information you need in order to make the decision that is right for you. They will also be able to help you discuss any emotional issues that may arise, and answer any questions or concerns you may have.



Remember that there is no going back after you receive your test results. It is

#### **d) Your Children's Risk**

##### **What the test results will mean for your children (and future children)**

The results of your genetic test will not only tell you about your risk of developing the condition, it will also tell you more about your children's risk.

If your test results show that you have not inherited the changed gene identified in your family, you are not at an increased risk of developing the condition, and your children will not be able to inherit the genetic risk from you. This is because you cannot pass on a changed gene that you do not have.

If your test results show that you do have the changed gene, your children might also have inherited the changed gene and be at risk. A child should not usually take a predictive test until they are over 18 years of age. If there is no medical benefit in testing a child, it is considered best to wait until the child is old enough to make the decision for him or herself. The exception to this is when there is a specific medical benefit in carrying out predictive testing on a child.

Discussing a genetic condition and predictive test results with children and adolescents can be very difficult. They may have lots of questions and it is important to answer any questions as honestly as possible taking into account their age and maturity level.



For some people, finding out they are at high risk of developing a condition is like being on an 'emotional rollercoaster'. They say there are good days and bad days. Most people do eventually come to terms with their result and use the information to help them make plans for the future.

### **How might I react if the result reveals that I do not carry the changed gene?**

For most people, finding out that you do not carry the changed gene brings joy and relief. However, it is not unusual for people to say that they experience a low feeling after testing. This may be because they have lived with the risk for so long that it requires a major change to adjust to being 'normal'. Some are disappointed that the 'good news' does not bring as many positive changes in life as they thought it would bring.

For some people who are convinced they do have the changed gene, finding out they do not can change their whole outlook on life. Some people find it difficult to deal with the idea of 'having a future'.

Some find it difficult to communicate the 'good news' to their relatives. 'Survivor guilt' is something that is often reported by people who receive 'good' test results. They wonder why they 'escaped' when other family members did not. Sometimes it is difficult to accept that you have been fortunate when others have not.

Some people who receive 'good news' feel an increased responsibility to care for family members who are affected by the condition.

It takes time to adjust to the results of the test, even if the results are 'good news'.

important therefore to try to think through some of the main issues before making a decision. Some of these are discussed below and might give you useful questions to think about and discuss. The list, however is not complete, and not all the points will be relevant to your specific situation.

## **Section 3. Making the Decision**

### ***a) Treatment and Prevention***

#### **Is there a way of treating or preventing the disease?**

It is important to find out whether there is a treatment available, or a way of reducing the risk of developing the condition, if you were found to have the changed gene. Knowing there is a treatment available might help in your decision making.

For some conditions such as the cardiac diseases mentioned, whilst there is no cure, the condition can be managed through drugs, specialised pacemakers or, in some cases, surgery.

For other conditions such as Huntington's disease, there is no treatment available to slow the progress of the disease but some of the symptoms can occasionally be partly managed with drugs. However, the benefit of knowing you have the changed gene may be more of an emotional benefit, or it may help when planning for the future.

### ***b) Uncertainty in Genetics***

#### **Uncertainty of the test results and the condition**

Predictive test results carry a degree of uncertainty, even if very slight. Having the changed gene usually means that it is almost certain that you will go on to develop the condition. However, it will still be difficult to predict when the condition will develop, how severe it will be or how rapidly the symptoms will progress.

Remember that, in scientific terms, a positive test result means that you DO carry the changed gene and a negative test result means that you DO NOT carry the changed gene. Most often the results will be one or the other.

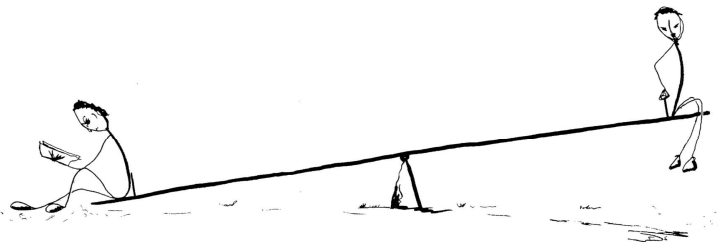
However, there is sometimes a third possibility: a grey zone or 'intermediate' test result. This means that, even though a changed gene is found, it is difficult to know whether in this case it will cause the condition. This is usually rare, but can be a very frustrating result to receive.

You should discuss all these possible outcomes with a genetic specialist when you are deciding whether to take the test.

### ***c) Dealing with the Results***

#### **How might the results of the test affect me emotionally?**

Before you make a decision about genetic testing, it is important that you try to think through, and discuss with a genetic health professional, how all the possible results of the test might affect you emotionally. It is a good idea to try to imagine how you might feel if you were to receive good news or bad news and recall how you reacted to bad news in the past. Thinking these issues through may help you in deciding whether it would be better to live with the uncertainty of not knowing or whether it would be better to know, whatever the result might be. However, it is important to remember that we all react differently and there is no 'normal' reaction.



#### **How might I react if the result reveals that I carry the changed gene?**

For some people, even a result which shows that they have the changed gene is preferable to the stress and anxiety caused by not knowing. For those people, having more 'information', whatever that information is, can be a relief.

Some people are relieved to find out they are at high risk of a genetic condition when it is something that can be treated. They feel that the information is useful as it means they can do everything they can to increase their chance of staying healthy. For others who have a changed gene for a condition that cannot be treated, it can be like finding out they have the condition already. The only question on their mind is 'when exactly will it happen to me?' This can be very distressing.

Some people experience a sense of shock when they find out they have the changed gene. They may feel alone, anxious, angry or ashamed. Genetic specialists and other health professionals, such as counsellors and psychologists, are experienced in helping people in these situations and can be a good source of support.

Some people also find it helpful to contact a patient association or support group. These groups can provide information about the condition and what it is like to live with it, including their experience with practical and emotional aspects. They can often put people and families in touch with others who are in a similar situation.

Finding out you have a changed gene and may have passed it on to your children can cause you to feel guilty and anxious about the future health of your children. It is important, however, to remember that genes are distributed by chance and having a changed gene is no one's fault.