

**Genetic Alliance UK**

Provides information about specific genetic conditions and contact details of support organisations.

Tel: 020 7704 3141

Email: [mail@geneticalliance.org.uk](mailto:mail@geneticalliance.org.uk)

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

**The Neurofibromatosis Association**

Tel: 020 8439 1234

Email: [nfa@zetnet.co.uk](mailto:nfa@zetnet.co.uk)

Web: [www.nfauk.org](http://www.nfauk.org)

**Genetics and Insurance**

More information about genetics and insurance can be found online at:

<http://www.abi.org.uk/BookShop/ResearchReports/Genetics%20Consumer%20Guide%20Low%20Res.pdf>



These contact details may no longer be correct.

Please check them online.

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This information was developed by Genetic Alliance UK, a national alliance of patient organisations which supports children, families and individuals affected by genetic disorders.

March 2009

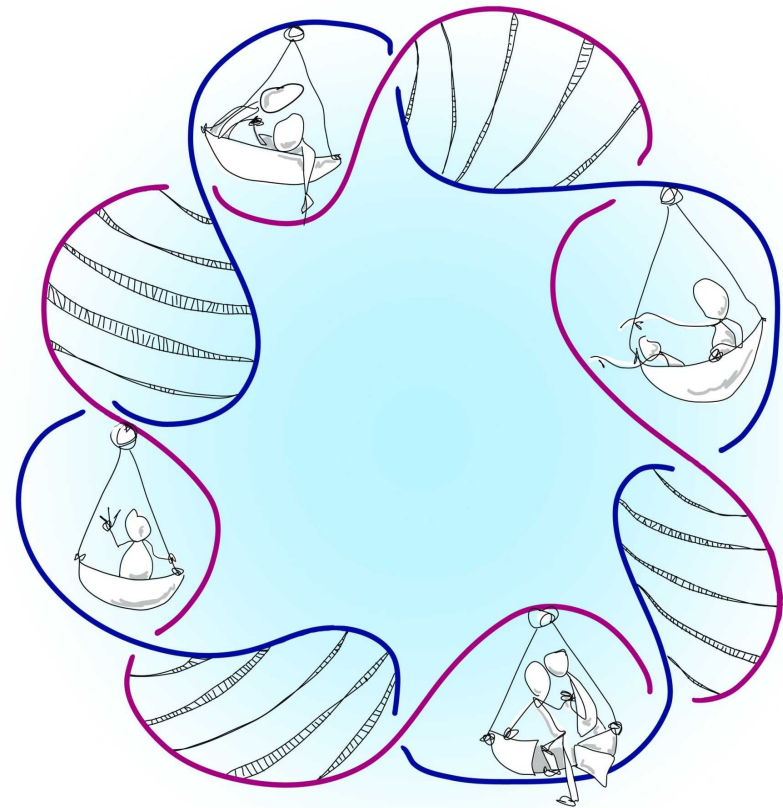
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Genetic Alliance UK  
 Supporting. Campaigning. Uniting.

# Predictive Testing for Inherited Cancer



## Information for Patients and Families

## Predictive Testing for Inherited Cancer

This information is about predictive genetic testing for inherited 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 why we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

Tel: 020 71216700

Web: [www.cancerresearchuk.org](http://www.cancerresearchuk.org)

**Macmillan Cancer Support (Cancerbackup)** – UK patient information group providing up-to-date cancer information, practical advice and support for cancer patients, their families and carers.

Tel: 0808 8001234 (specialist cancer nurse)

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

**AMEND – The Association for Multiple Endocrine Neoplasia Disorders**

Email: MEN1: [liz.dent@amend.org.uk](mailto:liz.dent@amend.org.uk)

Tel: 01423 780594 (9am-8pm only)

Email: MEN2/Sporadics: [jo.grey@amend.org.uk](mailto:jo.grey@amend.org.uk)

Tel: 01892 525308 (9am-8pm only)

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

**Childhood Eye Cancer Trust** - UK wide charity for families and individuals affected by retinoblastoma.

Tel: 020 73775578

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

**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)

**FAP Gene - Familial Adenomatous Polyposis**

Tel: 01664 566101

Email: [johnroberts@fapgene.org.uk](mailto:johnroberts@fapgene.org.uk)

Email: [mickmason@fapgene.org.uk](mailto:mickmason@fapgene.org.uk)

Web: [fapgene.org.uk](http://fapgene.org.uk)

### g) Timing of 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 e.g. I will definitely not do it until I am at 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:

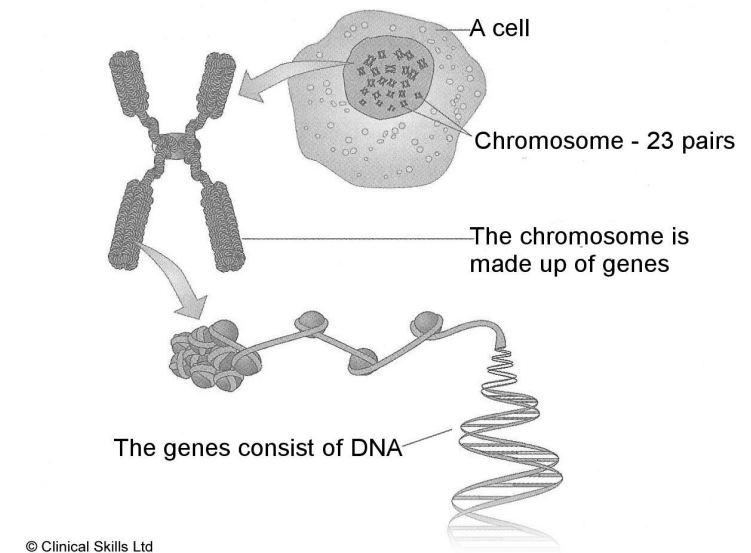
**Breakthrough Breast Cancer** - UK charity dedicated to fighting breast cancer through research, campaigning and education.

Tel: 08080 100 200

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

**Cancer Research UK** - Leading cancer charity dedicated to cancer research.

### Picture 1: Genes, chromosomes and DNA



Sometimes we inherit or develop a change (mutation) in a gene which stops it from working properly. This change can in some cases cause a susceptibility to certain cancers, because the gene is not giving the correct instructions to the body.

### Section 2. About the test

#### What is a predictive genetic test?

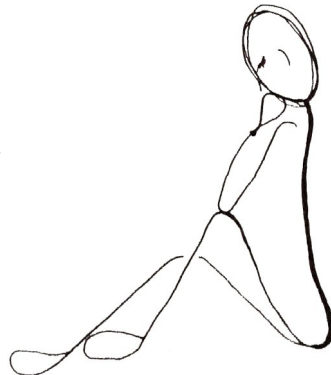
A predictive test (also known as a presymptomatic test) can provide information about whether or not a person is at an increased risk of developing a particular type of cancer. You can only take a predictive test if a close family member has had a genetic test which has identified a specific change in a gene that is associated with an inherited predisposition to cancer. This is because the cancer-causing gene first has to be identified in the family.

Whilst most cancers occur in adulthood, certain cancers may occur in childhood or adolescence (e.g., in individuals with multiple endocrine neoplasia or familial adenomatous polyposis). 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 the Genetics Laboratory?**' we provide more information about how genes are analysed.

### Why might I consider having predictive testing?

If there is a history of a particular type of cancer occurring in your family (usually two or more relatives from the same side of your family affected at a relatively young age e.g. under 60), it may be because of a changed gene in the family. Someone who has cancer at a particularly young age, or has had multiple cancers e.g. breast and ovarian cancer before the age of 50, is more likely than average to have a changed gene. If a changed gene has been detected in a close relative in your family, you might be able to have a predictive test to see if you have inherited this change. You might want to have a test if:

- That type of cancer can be effectively prevented or treated, or if there are screening measures that would be offered if you are at an increased risk.
- You believe that knowing more about your chance of getting that type of cancer may help you making important decisions about your life, including decisions about your health care (e.g. more regular check-ups or preventive surgery).
- You want the information to tell you more about the risk for your children.



## f) Confidentiality, Insurance and Finance

### Confidentiality

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

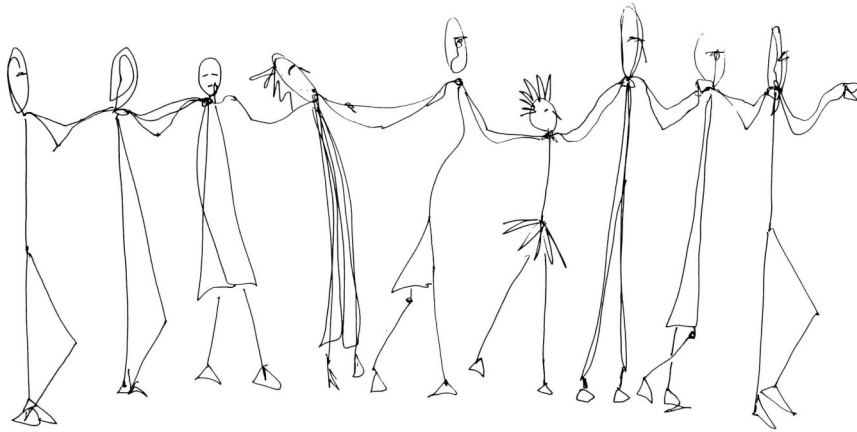
### Insurance

Insurance companies often ask you to provide medical details 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.





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.

- You are the type of person that prefers to know about your personal risk of developing the cancer as you prefer to know more about your future.

### Which types of cancer can be tested for by predictive testing?

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

- 1) Certain types of breast cancer and/or ovarian cancer.
- 2) Certain types of bowel, colon or womb (endometrial) cancer, including hereditary non-polyposis colorectal cancer (HNPCC) and familial adenomatous polyposis (FAP).
- 3) Other rare cancers, such as retinoblastoma, a rare cancer of the eye.

**Note: It is important to remember that cancer is a common condition which in most cases is caused by a combination of our genes, lifestyle and other environmental factors. Only a small minority, (5-10%) of cancers are inherited.**

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

For inherited cancers, having the changed gene means that you are at an increased risk of developing that particular type of cancer compared to people who do not have it in their families. For the vast majority of cases it does not mean that you will definitely get that type of cancer, but it increases the likelihood of developing it during your lifetime.

If you feel you may be at risk and want to consider having a predictive test for a particular type of cancer, you should make an appointment to see a genetic specialist. Before you decide about testing, you should ask the specialist to explain to you what your risk of developing the cancer will be if the test shows that you have the gene change.

## Genetic testing procedure

Before you can proceed with predictive testing, it must be confirmed that you are at risk and, if so, which gene change to look for if you do take the test. This is a 2-step process.



First, a family history will be done to assess the pattern of cancers in your family. A close relative of yours who is or has been affected by that particular type of cancer will then be tested in order to find the changed gene. If you have already had that type of cancer yourself, you could be the first person in the family to be tested (this type of test would then be known as a “diagnostic test” not a “predictive test”).

If a mutation is found in a relative then you can be offered a genetic test to see if you have inherited the changed gene. If no changed gene is found in a relative then it won't be possible to offer you a predictive test. This does not mean that you are not at an increased risk, in fact because of your family history you may still have a higher-than-average risk. Therefore you should still go for regular check-ups and screening.

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 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

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

The best way to avoid your children getting cancer is to make sure that they are:

- a) informed about the disease being in the family, and
- b) are aware of preventive measures available to help prevent that particular type of cancer.

For certain types of cancer, in particular the ones that affect children and adolescents, 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. However for the majority of cancers which occur in adulthood, continuously improving treatments and preventative measures are available, so prenatal testing is not often requested.

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 how the testing procedure, and the test results, might affect your relationship with your partner and other family members.



### **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 cancer, 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 have a 1 in 2 (50%) chance of also inheriting the changed gene and being at an increased risk of developing the cancer in their lifetime. 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, such as if screening for the cancer in your family is recommended for your child before the age of 18 years.

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.



you with all the information you need 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 important therefore to try and think through some of the main issues before making a decision. Some of these are discussed below and might give you some 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 a 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 way of reducing the risk of developing cancer, if you were found to have the changed gene. Knowing your risk reducing options beforehand may help you in your decision making.

For people who are at increased risk of developing cancer, frequent check-ups or screening can help pick up the condition during its early stages, meaning that treatment can be more effective. Some people may opt to have risk-reducing surgery. This means removing the organs (such as the breasts or ovaries) that are at increased risk of cancer.

**If you have a strong family history of a certain type of cancer that indicates that you may be at increased risk of developing it later on, the majority of management options to reduce that risk should still be available to you even if you do not have a genetic test. You should discuss this with your doctor.**

## ***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 will not tell you for certain whether you will go on to develop cancer, but will tell you that your risk is higher than for the average person in the population. If you do not carry the changed gene it does not mean that you will never develop cancer, but you have a similar risk to the general population.

## ***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 a good idea to try and 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. This 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. 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 persons, having more 'information', whatever that information is, can be a relief.

Some people are relieved when they find out they are at high risk for a particular type of cancer, when it is something that can be



treated. They feel the information is useful, as it means they can do everything they can to increase their chance of staying healthy. For others, finding out they are 'high risk' is like finding out they have the cancer 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 people to feel guilty and anxious about the future health of their children. It is important, however, to remember that genes are distributed by chance and having a changed gene is no one's fault.

### **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 a sense of relief. However some find it difficult to communicate the 'good news' to their relatives. 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.