

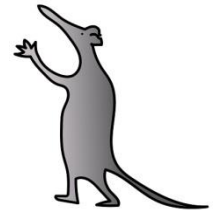
Haemophilia Centre

Genetic Testing and Consent for Patients and Families with Bleeding Disorders

Bleeding disorders may run in families and someone from your haemophilia centre will have explained to you how this affects your family. The purpose of this leaflet is to explain the reasons why you are being offered genetic tests and the consent form you will be asked to sign before these are performed.

Genetic tests may answer the following questions:

- If you are known to have a bleeding disorder, what is the genetic change that has caused your condition in your case?
- Are you a carrier of the bleeding disorder?



Introduction

Why do we resemble our parents? How does a single cell grow into a whole human? Genetics is the science that tries to answer these questions.

Humans, like every other living creature, are made up of cells. We all start off as one cell at the time of fertilisation. This cell contains two sets of genes, one from our mother and one from our father. For ease of storage and access, the genes are packaged up into 46 chromosomes. As the single cell divides the genes are copied so that every new cell possesses the full complement of genetic material. Genes are made of a chemical called DNA. Each cell holds about two metres of DNA (deoxyribonucleic acid).

Humans have approximately 30,000 genes stretched out along their DNA. Each gene acts as the recipe for the production of a protein and together they make up the recipe book, or blue print for you and me. Different genes, or recipes, are read at different times in different cells in response to the requirements of our bodies.

Sometimes genes, like recipes or blueprints, may have spelling mistakes in them or have bits missing. When this happens the proteins are either not produced or are abnormal. Genes, with these mistakes or mutations, function abnormally and so cause genetic disorders. Since genes are passed on from one generation to the next, genetic disorders often run in families. These mistakes can arise when a cell does not accurately copy its DNA. A mistake or variation in a single DNA letter can lead to disease.

Someone from your haemophilia centre has already explained the nature of the disorder and the manner in which it can be passed down through your family. If you require further information, or you are unclear about what you have been told, please ask for clarification or more help.

Genetic testing can tell us which people in your family have the condition and who are 'carriers' who might pass the disorder on to their own children. Simple tests of the defective clotting factor (coagulation factor) can sometimes tell us if a person is affected by the disorder or a carrier.



Sometimes the level is normal even though a person is carrying a defective gene. With modern genetic techniques it is usually possible to locate the faulty genetic change in each family, although this can sometimes take time. Although many families may have the disorder, it is common for each family to have its own unique genetic change.

What is the purpose of obtaining a blood sample?

It is very useful to know what the exact mistake in the DNA is that is causing the disorder in you/your child. Sometimes this helps us to be warned about how the disorder may respond to treatment in the future. Measurement of the blood coagulation factor level does not always clearly indicate if there is a genetic mistake present or not. Analysis of the DNA is a more accurate way of telling this. For this, a special type of blood sample is required from which the DNA can be extracted. A second sample may be taken from you on a separate occasion to confirm the result of the initial test.

Where will the blood sample be tested?

The tests needed to detect a change ('spelling mistake' or bit missing) in DNA are specialised. Some of them are performed locally, but depending upon the nature of the disorder, it may be necessary to send your blood sample away to one of a small number of specialised laboratories. In all these, there are strict regulations in place to ensure complete confidentiality of your details.

How long will the test take?

The answers to genetic tests often take some time to obtain. Your doctor will discuss the likely time course with you, as this varies with the disorder. It may take several months, or years if you have one of the less common, or more complicated disorders.

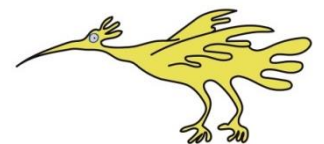
How long will my blood sample be stored?

Sometimes it may not be possible with existing methods to find the genetic change in your family. In this case, the DNA will be stored until new tests are available. It is usual practice to store DNA samples indefinitely. Other new tests relevant to the disorder may arise in future, which will help us understand more about the mechanisms of the disorder.

What are the risks of genetic testing?

In addition to information on the inheritance of a bleeding disorder, the results from these genetic tests are likely to be able to determine other information, such as confirmation of whether a child's parent is as assumed by the family. Therefore, occasionally unexpected results about family relationships arise from these tests, which, if known, could cause embarrassment within a family. If it is found, for example, that an individual's parent is different from that assumed by the family, significant psychological problems can be caused, and this may cause harm to the person being tested and other family members.

The studies performed will be specific for the disorder known to be in your family. They will not exclude all forms of possible bleeding disorders.



What else might be done with my blood sample?

We might want to use your sample to help develop or refine tests for bleeding disorders. In such cases your blood samples would not be linked back to you. The results would therefore be completely anonymous. From time to time it is very useful to run tests on a series of DNA samples anonymously to compare how common some changes in the DNA are which are not responsible for the condition. If your sample is used for such testing, no one will know whose it is, and there will be no comeback to you and your family.

Who gets to know about the results?

The results will be told to you personally. Your family doctor will be sent the result.

Why might it be useful for other members of my family to know about the results?

Information about the genetic disorder in you/your child is likely to be of benefit to other members of your family. It may, for example, be used to discover if a woman is a carrier and therefore if there is a risk of passing on the disorder to her children. With your permission, we would like to be able to make the information about your genetic change available to doctors looking after other people in your family if they ask.

Are my genetic results going to be stored anywhere other than in my hospital and GP case records?

There are local and national confidential databases, which keep information about genetic disorders of coagulation. We would like to record the information about your gene change. These databases are secure and protected.

If you would like to have your blood tested please read the consent form on the next page.

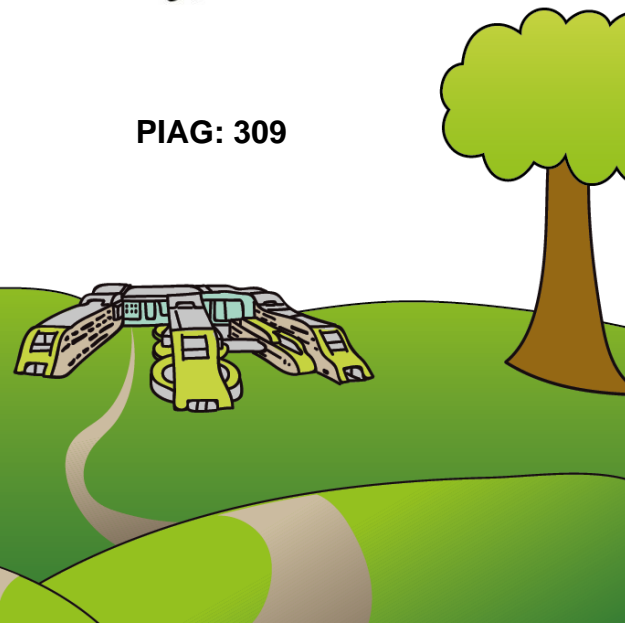
The content of this leaflet and consent form was developed by the United Kingdom Haemophilia Centre Doctors' Organisation (UKHCDO) <http://www.ukhcdo.org/>.

This leaflet only gives general information. You must always discuss the individual treatment of your child with the appropriate member of staff. Do not rely on this leaflet alone for information about your child's treatment.

This information can be made available in other languages and formats if requested.

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Consent for a Genomic Test

I agree for a sample from myself / my child / my deceased relative to be tested to look for genetic changes potentially linked to:

Name of Clinical Indication	
Testing for Familial Variant	<input type="checkbox"/> Y / <input type="checkbox"/> N

Name of person whose sample is to be tested		Date of Birth	
NHS Number		Hospital Number	

Possible results:

- A change may be found that is the whole or at least partial cause of the condition. It may have other health implications for me and / or other family members.
- No differences may be found i.e. testing may not find the cause of the condition.
- A change may be found that cannot be interpreted with current scientific knowledge (further family studies or research may be helpful).
- In rare situations a change may be found that is not thought to be the cause of the condition being investigated, but could have other implications for me and / or other family members.

I agree to the following:

	Please Initial
Use of the test results by health care professionals to help other family members	
Storage of the sample and / or genomic data in case future relevant tests become available.	
Signature	Date

If signing on behalf of your child or deceased relative, please also indicate:

Your Name	Relationship

Child's assent to genomic testing:

I understand the reason for this test: Y / N I agree to this test: Y / N

Your Name	Signature	Date

Name of the clinician taking the consent	Signature	Date
Designation of clinician:		