East Kent Hospitals University NHS Foundation Trust

Genetic testing for inherited bleeding disorders

Information for patients and relatives from the Kent Haemophilia and Thrombosis Centre

Bleeding disorders may run in families, and someone from the Haemophilia Centre based at Kent and Canterbury Hospital will have explained to you how this affects your family. The purpose of this information sheet is to explain the reasons why you are being offered genetic tests, and why you will be asked to sign a consent form before these are performed.

Genetic tests may answer the following questions.

1. If you are known to have a bleeding disorder, what is the genetic chance that has caused your condition in your case?

2. Are you a carrier of the bleeding disorder?

What are genes?

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 in to 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 Deoxyribonucleic acid (DNA). Each cell holds two metres of DNA. 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 and different cells in response to the needs of our bodies.

What are genetic disorders?

Sometimes genes, like recipes or blueprints, may have spelling mistakes in them or bits missing. When this happens the proteins are either not produced or are abnormal. Genes with these mistakes or mutations do not work normally and cause genetic disorders. As 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 DNA. A mistake or variation in a single DNA letter can lead to a disease.

Someone from the Haemophilia Centre will have already explained the nature of your disorder, and the manner in which it can be passed down through your family. If you need further information or you are unclear about what you have been told, please ask a member of staff.

What can genetic testing tell me and my family?

Genetic testing can tell you which members of 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 is a carrier.

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

Why collect a blood sample?

It is very useful to know what the exact mistake in the DNA is. 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 (FVIII or FIX for example) does not always clearly show 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 needed from which the DNA can be taken. A second sample may be taken from you at a separate time to confirm the result of your first test.

The blood sample will be taken from your arm; the area at the bend in your elbow in the front of your arm (known as the anti cubital fossa) is usually used. If access to this vein is difficult, the back of your hand may be used instead.

Is there another way of finding this out?

No, there is currently no other way.

Where will the blood sample be tested?

The tests needed to detect a change in DNA are specialised. Some tests are done locally, but depending on the nature of your disorder, it may be necessary to send your blood sample away to a specialised laboratory.

In all these laboratories there are strict regulations in place to make sure complete confidentiality of your details.

How long will the test take?

The answer to genetic tests often takes some time. Depending on the type of disorder you have and the purpose of the test, it may be weeks, months, or some times a year, if you have one of the less common or complicated disorders. Your doctor may be able to give you an estimate of the time it will take.

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. If this happens your DNA will be stored until new tests are available. It is usual practice to store DNA samples indefinitely. Other new tests may be available in the future, which will help us understand more about the mechanisms of your disorder.

What are the risks of genetic testing?

In addition to information on the inheritance of a bleeding disorder, the results from these tests may be able to determine information such as confirmation of parentage. Occasionally unexpected results about family relationships arise from these tests, which if known could cause embarrassment within the family. If it is found for example that an individuals parent is different from that assumed by the family, significant psychological problems can be caused and this may result in harm to the person being tested and other family members. If you have concerns about this, please discuss them with your healthcare professionals before testing begins. This information will remain confidential between you and your healthcare provider.

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 are the benefits to having a test done?

Genetic tests can help us plan the future management of your condition, and allow you to make choices when preparing for your future.

What else might be done with my blood sample?

When your tests are completed, any remaining DNA will be stored. It may be used:

- for further testing if new tests for your condition become available
- as a sample to be compared with other samples taken from family members and unrelated individuals who have the same condition; or
- 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 be completely anonymous.

Who gets to know about my results?

The results will be told to you personally. Your family doctor will also be sent your results.

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

Information about the genetic disorder in you or 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 here. These databases are secure and protected.

Contact details

If you have any further questions or concerns, please contact the Kent Haemophilia and Thrombosis Centre on 01227 783157.

If you would like to have your blood or your child's blood tested, please read the consent form.

This leaflet has been produced with and for patients.

Please let us know:

- If you have any accessibility needs; this includes needing a hearing loop or wanting someone to come with you to your appointment.
- If you need an interpreter.
- If you need this information in another format (such as Braille, audio, large print or Easy Read).

You can let us know this by:

- Visiting the Trust web site (https://www.ekhuft.nhs.uk/ais).
- Calling the number at the top of your appointment letter.
- Adding this information to the Patient Portal (https://pp.ekhuft.nhs.uk/login).
- Telling a member of staff at your next appointment.

Any complaints, comments, concerns or compliments, please speak to a member of your healthcare team. Or contact the Patient Advice and Liaison Service on 01227 783145 or email (ekh-tr.pals@nhs.net).

Patients should not bring large sums of money or valuables into hospital. Please note that East Kent Hospitals accepts no responsibility for the loss or damage to personal property, unless the property has been handed into Trust staff for safe-keeping.

Further patient information leaflets are available via the East Kent Hospitals' web site (https://www.ekhuft.nhs.uk/patient-information).

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