

Introduction

The purpose of this information sheet is to explain the reasons why you are being offered genetic tests for thrombophilia. You will be asked to sign a consent form before these tests are performed. Genetic tests may help to answer the following question: Do you have an inherited trait which may make you more likely to develop a clot in the future?

Genetics

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. Each cell in the body contains two sets of genes, one from our mother and one from our father. Genes are made of a chemical called 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. Sometimes genes, like recipes or blueprints, may have spelling mistakes in their DNA or have bits missing. When this happens the proteins are either not produced or are abnormal. Genes with these mistakes called “mutations” function abnormally and cause genetic disorders. Since genes are passed on from one generation to the next, genetic disorders often run in families.

Genetic testing can tell us which people in your family have the condition and who might pass the disorder on to their own children.

What is the purpose of obtaining a blood sample?

It is very useful to know what the exact mistake in the DNA of the gene as this information may help your doctor to decide whether you have higher chance of developing a clot in the future and may be used for the investigation of other family members.

Where will the blood sample be tested?

The blood sample will be tested in the National Coagulation Laboratory at St. James’s Hospital. There are strict regulations in place to ensure complete confidentiality of your details.

How long will the test take?

The result should be available within two months.

How long will my blood sample be stored?

Blood samples are stored for a maximum of two years.

What are the risks of genetic testing?

In some cases, identifying the faulty gene might affect individuals with regard to insurance. Advice from an insurance broker should be considered if necessary.

What else might be done with my blood sample?

With your consent the blood sample may be used anonymously for quality assurance, research or development of new tests.

Who gets to know about the results?

The results will be sent to your doctor who has ordered the test who will then give you the results.

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

Information about the genetic disorder in you is likely to be of benefit to other members of your extended family in the future. With your consent your clinical and genetic information may be made available to relevant healthcare professionals.

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

The information will be stored in a local confidential database, which keeps information about genetic disorders of coagulation. These databases are secure and protected.



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