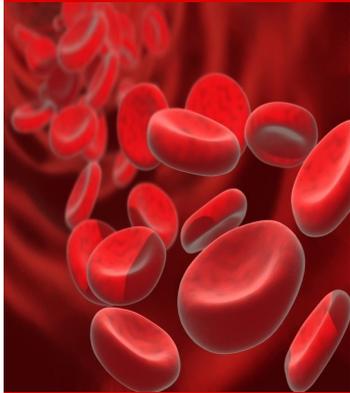


# Genetic Testing for Haematological Disorders

## Patient Information Leaflet



## Beaumont Hospital Haematology Laboratory



Ref: LP-GEN-0014,16

### ***Introduction***

The Purpose of this information sheet is to explain the reason why you are being offered genetic testing. You will be asked to sign a consent form before these tests are performed. Genetic tests may help to answer the following questions: Do you have an inherited or acquired trait which may make you more likely to develop this condition. Identifying this trait may help your doctor in your diagnosis and treatment.

### ***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 nucleated cell in the body contains two sets of genes, one from the father and one from the mother. 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 may cause genetic disorders.

### ***What is the purpose of obtaining a blood sample?***

It is very important to know the exact mistake or “mutation” in the DNA of the gene as this information may help the doctor to diagnose your disorder or decide the risk of developing a disorder in the future. It may also be used for the investigation of other family members if deemed necessary by your doctor.

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### ***Where will the blood sample be tested and how long will the test take?***

The blood sample will be tested in the Haematology Laboratory in Beaumont Hospital. There are strict regulations in place to ensure complete confidentiality of your details. The results should be available within two months.

### ***How long will my blood be stored?***

Blood samples requiring testing for inherited Thrombophilia disorders are stored for a maximum of 6 months. Blood samples requiring testing for Haematological malignant disorders are stored indefinitely.

### ***What are the risks of genetic testing?***

In some cases, identifying the faulty gene might affect individuals with regards to insurance. Advice from a clinician 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 the results?***

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

### ***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. This database is secure and protected.

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