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Central Manchester University Hospitals NHS
NHS Foundation Trust

Manchester Royal Infirmary

Information on Genetic Testing and Consent Form for Patients and Families with Bleeding Disorders

Comprehensive Care Haemophilia Centre

INVESTORS IN PEOPLE

POSITIVE ABOUT DISABLED PEOPLE

Introduction

The purpose of this information sheet 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.

Someone from your haemophilia centre has already explained the nature of your disorder to you, 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. This can be more helpful than simple tests of the defective clotting factor (coagulation factor) where sometimes the level is normal in carriers. With modern genetic techniques it is 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.

1. What is the purpose of obtaining a blood sample?

It is very useful to know exactly the genetic change that is causing the disorder in you/your child. Sometimes this helps us to be alerted 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 change present or not; genetic testing is a more accurate way if telling this. For this a special type of blood sample is required from which the genetic material (DNA) can be extracted.
A second sample may be taken from you on a separate occasion to confirm the result of the initial test.

2. Where will the blood sample be tested?

The tests needed to detect a genetic change are specialised. Some of them are performed locally, but depending upon the nature of your disorder, it may be necessary to send you blood sample away to one of a small number of specialised laboratories. In all these, there are strict regulations to ensure complete confidentiality of you details.

3. How long will the test take?

The answer 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 many months if you have one of the less common, or more complicated disorders. You will be informed of progress if it will take a long time to obtain results.

4. How long will my blood sample be stored?

Sometimes it may not be possible with existing methods to find 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 your disorder may arise in future, which will help us understand more about the mechanisms of your disorder.

5. What are the risks of genetic testing?

In addition to information on the inheritance of a bleeding disorder, the results from these genetic tests may inadvertently provide 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 unknown, could cause embarrassment or upset within a family.

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.

6. 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. It can be 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.

7. Who gets to know about the results?

The results will be told to you personally. Your family Doctor will be sent the result, unless you withhold consent for this.

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

Information about genetic change 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 women 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.

9. Who should give consent for testing a child?

A child may not provide informed consent until he/she is mature enough to understand the implications of the test being performed. Genetic carrier testing will not normally be carried out before a child is mature enough to understand for themselves the implications of the test results. Information about the genetic change in a child affected by the bleeding disorder may affect treatment and is likely to be of benefit to other family members. In this case the parent or legal guardian of the child will be asked to provide informed consent.

10. 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 and comply with Caldicott and the Data Protection Act.

Further information on general issues of consent can be found in the Trust's "Confidential to Treatment" leaflets for patients and parents. Please ask for a copy if these have not been provided to you.

If you would like to have your blood tested please read the attached consent form.

Background information

Why may we resemble our parents? How does a single cell become 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 form 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 (deoxyribonucleic acid).

Humans have approximately 30,000 genes stretched v 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 requirement 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 can lead to disease.

No Smoking Policy

The NHS has a responsibility for the nation's health.

Protect yourself, patients, visitors and staff by adhering to our no smoking policy. Smoking is not permitted within any of our hospital buildings or grounds.

The Manchester Stop Smoking Service can be contacted on Tel: (0161) 205 5998 (www.stopsmokingmanchester.co.uk).

Translation and Interpretation Service

It is our policy that family, relatives or friends cannot interpret for patients. Should you require an interpreter ask a member of staff to arrange it for you.

تنص سياستنا على عدم السماح لافراد عائلة المرضى او اقاربهم او اصدقائهم بالترجمة لهم. اذا احتجت الى مترجم فيرجى ان تطلب ذلك من احد العاملين ليرتب لك ذلك.

ہماری یہ پالیسی ہے کہ خاندان ، رشتہ دار اور دوست مریضوں کےلئے ترجمہ نہیں کرسکتے۔ اگرآپ کومترجم کی ضرورت ہےتو عملے کےکسی رُکن سے کہیں کہ وہ آپ کےلئے اس کا بندوبست کردے۔

ইহা আমাদের নীতি যে, একজন রোগীর জন্য তার পরিবারের সদস্য, আত্মীয় বা কোন বন্ধু অনুবাদক হতে পারবেন না। আপনার একজন অনুবাদকের প্রয়োজন হলে তা একজন কর্মচারীকে জানান অনুবাদকের ব্যবস্থা করার জন্য।

Nasze zasady nie pozwalają na korzystanie z pomocy członków rodzin pacjentów, ich przyjaciół lub ich krewnych jako tłumaczy. Jeśli potrzebują Państwo tłumacza, prosimy o kontakt z członkiem personelu, który zorganizuje go dla Państwa.

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我们的方针是，家属，亲戚和朋友不能为病人做口译。如果您需要口译员， 请叫员工给您安排。

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