

Appendix I

This space is for heading for hospital and Haemophilia Centre details.) A word version of this information sheet and consent form for use (and if appropriate modification) is available from:

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Information on Genetic Testing and Consent Form for Patients and Families with Bleeding, Thrombotic and /or Platelet Disorders

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, 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. Although many families may have the same 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 DNA 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 levels of blood coagulation factors or platelet function do not always indicate if there is a DNA change present or not; genetic testing is a more accurate way of telling this. For this a blood sample is required from which the DNA (genetic material) can be extracted. On occasions a second sample may be taken from you to confirm the result of the initial test.

The genetic test will include a comprehensive screen of about 100 different genes (you have ~21,000 genes in your DNA). Only changes in the gene(s) relevant to the disorder being tested for will be reported.

- 2. Where will the blood sample be tested?** In a laboratory that is part of the NHS or affiliated to the National Institute for Health Research. There are strict regulations in place in the testing laboratories to ensure complete confidentiality of your personal details such as surname, first name and date of birth.
- 3. How long will the test take?** The genetic tests generally take some months before they are completed. You will be informed if it will take more than three months to obtain results.
- 4. How long will my blood sample be stored?** Sometimes it may not be possible with the existing genetic tests to detect the DNA change in your family. In this case, the DNA will be stored for possible future use when better tests have become available; it is usual practice to store DNA samples indefinitely. New tests may be developed in the future which will help us understand more about the genetic cause of your disorder.
- 5. What are the risks of genetic testing?** In addition to information on the inheritance of a bleeding, thrombotic or platelet disorder, the results from these genetic tests may inadvertently provide other information about family relationships, such as paternity. If the result is unexpected, this, may cause embarrassment or upset within a family.

The tests that will be performed will include all genes known to underlie bleeding, thrombotic or platelet disorders. This test will not detect genetic abnormalities that are associated with other unrelated disorders. Currently the number of genes included in the test is close to one hundred; the gene content of the test will be reviewed at regular intervals so that new more recently discovered genes can be included.

6. **What else might be done with my blood sample?** We might want to use your blood or DNA sample to help develop tests for bleeding, thrombotic or platelet disorders. It can be very useful to run tests on a series of samples anonymously to compare how a new test compares to the current ones. If your sample is used for such testing, no one will know whose it is; the results would be completely anonymous. Therefore there will be no comeback to you and your family.
7. **Who gets to know about the results?** The laboratories which perform the tests on your blood or DNA will return the results to the Haemophilia centre and a member of your clinical care team will inform you about the results personally. Your family doctor will be sent the result by the clinical care team at your Haemophilia centre, 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 the 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 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.
9. **Who should give consent for testing a child?** A child may not provide informed consent until he or she is mature enough to understand the implications of the test being performed. This age varies with the individual child. Genetic carrier testing will not normally be carried out before a child is of an age to appreciate the issues and give consent. However, there may be reasons why the results of such a test would be valuable. Information about the genetic change in a child affected by the disorder may affect treatment and is likely to be of benefit to other family members. In both of these cases 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, national and international anonymised databases, which keep information about DNA variants linked with bleeding, thrombotic and platelet disorders. Your surname, first name and date of birth will never be made available to such databases but the DNA variant observed in you/your child linked with your clinical symptoms and laboratory results will be released. The sharing of these data in an anonymised manner will lead to continued improvements in the accuracy with which genetic tests can be reported.
11. **What will happen if I decide to withhold consent?** You may withhold consent for any or all of the above uses for samples and results. This would not jeopardise your treatment (or that of your child).

Further information on general issues of consent can be found in the Trust's "Consent 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 complete the attached consent form.

A. Patient details

Surname:	Consultant:
Forename:	Hospital Number:
Date of birth:	

B. Collection and usage of samples

I, (print name) give consent for a blood sample to be taken from (myself or name of child) and the genetic material extracted, stored and tested for (specify disorder).

Please initial the boxes below to indicate your consent

- The purposes for obtaining this sample and the potential implications have been explained to me and I have had an opportunity to have my questions answered.
- I have read and understood the above information about genetic testing.
- It is the intention to store the sample indefinitely.
- If no relevant test is currently available, I agree to the sample being stored until such time as an appropriate test is developed and the sample may then be tested.
- I understand that it may be necessary to use part of the sample anonymously for example for quality assurance or development of new tests.

Signed: Date:

Patient/parent/legal guardian
delete as appropriate

C. Use and availability of results

- I hereby give consent for clinical, laboratory and genetic information that may be relevant to other family members to be made available to relevant health care professionals.
- I agree to the results being entered into local and national confidential databases
- I agree that the results of the genetic test together with my clinical symptoms and laboratory results will be released in publically accessible anonymised databases for use by others to improve the reporting of genetic test results.

Signed: Date:

Patient/parent/legal guardian
delete as appropriate

D. Person obtaining consent

I have explained to the above patient/parent/legal guardian the purpose of obtaining a sample for genetic studies and their implications.

Signed: Date:

Print name: Position:

A photocopy of the completed form should be given to the patient, the original filed in the patient's case notes and a copy filed in the family genetic record file.