West Midlands Familial Hypercholesterolaemia Service

Cascade Family Testing Patient Information Leaflet

This leaflet aims to explain what Familial Hypercholesterolaemia is and why this service may be important to you and your family.

How we identify family members who may be at risk of Familial Hypercholesterolaemia (FH)?

Family testing is recommended for all blood relatives of someone found to have FH and you have received this leaflet because a member of your family has been confirmed as having FH. This means that you have a 50% chance of inheriting this condition. We therefore recommend that you have an FH genetic test.

What is Familial Hypercholesterolaemia (FH)?

Genes carry information that determine who you are. FH is caused by an alteration in a gene that is responsible for helping your body clear cholesterol from the blood. High blood cholesterol levels can lead to blockages in your arteries and can cause conditions like heart disease. Early diagnosis of FH and treatment can reduce the risk of this happening. Children have a 50% chance of inheriting FH if their parents have the condition.

FH treatment and how it can help you?

FH is a treatable condition, and the earlier treatment is started, the more effective it will be. The treatment (medication) reduces the risk of developing early heart disease.

FH is diagnosed by taking a sample of blood or saliva (spit) which is sent to a laboratory for genetic testing. If you are found to have inherited the same gene alteration found in other members of your family, other close relatives including children, can also be offered an FH test.

It is important to identify FH in childhood, particularly in families where heart disease is occurring in early adult life.

What are genes and how are they involved in FH?

Genes are coded messages and contain instructions for how cells in our body grow and function. If there is a change or an alteration in one of the genes that control cholesterol, that gene won't function correctly, and your cholesterol level will be higher. If one of these

gene alterations has been identified in a family member, other family members who are at risk of having FH should be tested for that specific gene alteration.

What if I know my cholesterol isn't high or if I've never had it checked?

Depending on the genetic alteration that is passed down in the family, occasionally a person with FH can still have 'normal' cholesterol levels, or levels that are only slightly higher than 'normal'. Even if your cholesterol levels are 'normal', if FH has been identified in your family, it is still important to have a genetic test. Not being tested may leave this condition undiagnosed, untreated and could increase your risk of developing early heart disease.

What will happen at my FH clinic appointment?

During your appointment, you will be asked about your medical history, as well as the medical history of your family. Most appointments can be done via telephone with a specialist nurse. The nurse will explain FH, the testing process and answer any questions you may have. After the appointment, a saliva (spit) sampling kit will be sent to you at home.

There are a small number of FH gene alterations that require a blood sample for testing. If this is the case, you will be offered a face-to-face appointment with a specialist nurse for a blood sample to be taken. If you are unable to attend a face-to-face appointment, we can still arrange a telephone appointment and discuss the process for having your blood sample taken at a local phlebotomy service. When obtaining any sample for genetic testing, your written consent will be required and the FH nurse will discuss the consent form with you during your appointment.

Your sample

Your sample will be sent to a laboratory to see if you have inherited FH. It takes up to four weeks to receive the results of the test and you will be contacted by letter or telephone with the results.

You will receive a copy of your genetic result and your GP will also be notified. If your test is positive (confirming that you have inherited Familial Hypercholesterolaemia) we will contact you via telephone to discuss the result. We will also review your family history again and discuss who else in the family should be tested for FH.

With your consent, we will refer you to a specialist consultant who will support you to manage your FH. With your agreement, your sample will be stored at the specialist genetics laboratory that tested your sample, and normal laboratory practice is to store the DNA extracted from the sample even after the current testing is complete. During your appointment you will be asked to complete the consent form.

Protection of information and confidentiality

All the information you give to us will be kept private and it will stay confidential. Your information will be stored on a secure electronic database, which is an FH register. This allows us to co-ordinate family members who may also be tested for and diagnosed with FH. All information obtained by our service is protected and governed by the General Data Protection Regulation (GDPR) in conjunction with the Data Protection Act 2018. In addition, all staff must comply with the Common Law Duty of Confidentiality and various national and professional standards and requirements.

Any questions or concerns?

If you have any questions or concerns about this information or the service you are being offered, please contact the FH team on **0121 371 8179**. If after speaking with one of the FH team you have concerns that you feel haven't been answered, you can contact the Patient Relations Department at University Hospitals Birmingham NHS Foundation Trust on **0121 424 0808** or email: **PALS@uhb.nhs.uk**

Further information regarding cholesterol and FH can be found on the following websites: **www.bhf.org.uk, www.heartuk.org.uk**

How did we do? ☺ 😐 ☹

If you have recently used our services we'd love to hear about your experience. Please scan the QR code or follow the link to share your feedback to help us improve our services. **Thank you. www.uhb.nhs.uk/fft**

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