West Midlands Familial Hypercholesterolaemia Service

Patient Information Leaflet

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

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

If someone is identified as being at risk of FH, such as patients with high cholesterol levels, they will be invited to attend an appointment with a specialist nurse at a local GP surgery.

What is Familial Hypercholesterolaemia (FH)?

FH is an inherited condition. People with FH have high cholesterol levels from birth which means they are at greater risk of developing early heart disease. FH is caused by an alteration in a gene which is responsible for helping your body clear cholesterol from the blood. High blood cholesterol levels can lead to partial or total blockages of the arteries causing heart disease, however, early diagnosis and treatment can reduce the risk of this happening. Children have a 50 per cent chance of inheriting FH if their parents have this 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 in preventing early heart disease. FH is diagnosed by taking a sample of blood which is sent for genetic testing. If you are found to have the gene, other close members of your family, 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 which give instructions for how cells in our body grow and function. Genes come in pairs and we inherit one copy from each of our parents. We know that FH can be caused by changes in four genes –

- the LDLR gene which helps to remove cholesterol from the blood
- the APOB gene that holds the cholesterol particles together in the blood
- the PCSK9 gene which regulates the removal of cholesterol by the LDLR gene

- the APOE gene which also helps to remove cholesterol from the blood

An alteration in one of these genes leads to increased levels of cholesterol causing a higher risk of heart disease. It is believed that there may be other genes yet to be identified that cause FH. It is possible that further tests may become available in the future.

What will happen at my clinic appointment?

You will be seen by an FH specialist nurse who will answer any questions you might have. You will be asked about your medical history as well as the medical history of your family. Following the initial assessment, it may be necessary to take a blood sample to send for genetic testing which will need your written consent.

Your blood sample

Your blood sample will be sent to a genetics laboratory for analysis to see if you have FH. The FH nurse will explain the process in more detail at your appointment.

It takes up to eight weeks to receive the results of the blood test and you will be contacted by letter with the results. You will also receive a copy of your blood results and your GP will also be notified.

If your test is positive you will be invited to attend a follow up appointment to discuss the results. We will also ask you to complete a family history form and bring this with you to your follow up appointment. We suggest that you discuss this with your family members in case we need to invite them for screening.

With your agreement your blood sample will be stored at the Bristol Genetics laboratory and normal laboratory practice is to store the DNA extracted from the blood sample even after the current testing is complete. The reason for this is that in the future (months or years) further/new tests may become available. We will discuss this with you in clinic and complete the consent form accordingly.

In some instances, leftover samples may be useful in checking laboratory techniques and there are occasions when your sample might be used as a ‘quality control’ for other testing. We will discuss your blood test in detail at your appointment.

Protection of information and confidentiality

All the information we obtain about you will be strictly confidential. Your information will be stored on a secure electronic database which is a FH register and allows national coordination of family members who may be diagnosed with FH. All information obtained by the FH 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.

Insurance policies

The following organisations have more detailed information and details on insurance issues.

www.bhf.org.uk

www.heartuk.org.uk

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 371 4400 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

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UHB PI18/1941/06 Author: Elaine George Date: June 2020 Review Date: June 2022