All Wales FH Family Forum

- for individuals and families with inherited high cholesterol.

Research and Development - Special Edition 2014

Service Developments
Extending our reach

Helping your doctor to help you

Raising awareness of FH is a key aim of the FH team. General Practitioners (GPs) often don't have resources to keep up to date with the latest developments. The FH team has developed a web-based teaching package on FH for GPs and Practice Nurses that will help practices understand more about FH. The FH nurses are also helping GPs and cardiologists by looking for patients on their lists who may have FH.

FH is a family affair – A reminder from Dr Peter Dale (Paediatrician)

Family cascade testing from parents and grandparents who have an FH gene is important so that children and young people who inherit the gene can be recognised early and offered treatment when appropriate. Genetic testing in children is very easily carried out by either blood or saliva samples when the family gene is known. We recommend that children are genetically tested from the age of eight years and offered treatment by the age of 10.

Every family should participate in cascade testing so that their children and grandchildren have the best chance of growing up to be healthy young adults without the increased risks of cardiovascular disease.

Watch out -

A new cartoon for children with FH will be posted on the FH Wales website soon.

Statins – reported side effects exaggerated

Most people who take statins feel completely well and have no side effects. There is strong evidence that they reduce the risk of heart disease in FH and overall the safety record of statins is very good. A few patients report muscle aches and pains when they are taking statins. These effects are reversible and it is generally possible to find a type or dose of statin that is suitable, or an alternative medication. There may also be a slight increase in blood sugar and, for a few people, this may move them from a state of borderline diabetes into the diabetic range.

There has been recent debate about this topic in the British Medical Journal who published an article stating that 20% of patients taking statins would develop side effects. However, the authors subsequently withdrew that figure because the data was flawed and the BMJ published a statement to say that the risks had been overestimated. The National Institute for Clinical Excellence (NICE) recommends statins as the first line treatment for FH because the cholesterol lowering benefits far outweigh the risks for FH patients. If you have any questions about this issue it is best to discuss with your doctor or FH nurse.

LDL- Cholesterol Apheresis

Most patients with FH respond well to treatment with statins and other lipid lowering medications. A very small percentage of FH patients, however, have persistently high cholesterol levels despite taking maximum doses of these treatments. This may put them at risk of heart disease. For these patients LDL- cholesterol apheresis is a 'lifeline'. Apheresis is a procedure which is like kidney dialysis - the patient's blood is passed through a column to remove LDL-cholesterol and their 'cleaned blood' is then returned back to them. This procedure takes about two hours and needs to be repeated every two weeks. For patients, this procedure requires great commitment, but provides long term health benefits. There are only seven apheresis units in the UK and we are very fortunate in Wales to have one located at Llandough Hospital, near Cardiff. Last year a patient evaluation of the service rated it 10 out of 10. Eighteen FH patients regularly have apheresis at the Llandough Lipid Unit, some for over 20 years, and are cared for by the nursing team led by Specialist Nurse Suzanne Watkins under the medical direction of Dr Dev Datta.

www.cardiffandvaleuhb.wales.nhs.uk/lipidunit

FH testing – England following Wales' lead

The All Wales FH Cascade Testing Service has led the way on FH services in the UK. The FH Family Forum has given lots of support to Heart UK and the British Heart Foundation in their efforts to lobby for a similar service in England. At last, progress is being made! The British Heart Foundation, an important supporter of the All Wales Service, has recently announced that it will fund FH Specialist Nurses across England.

www.bbc.co.uk/news/health-27586009

Dr Ian McDowell from the Wales FH service has been asked to contribute to the NHS England FH steering team to share our experience from Wales. The English centres will be using a patient management system similar to that used in Wales. Kate Haralambos from our Wales FH team has been asked to help support the development of this system in England.

Research News

Introducing the Research Team

We are always seeking to develop our service and learn more about how best to diagnose and treat FH. A range of people are involved with research into various aspects of FH and service development including nurses, consultants, clinical scientists and geneticists. Currently two dedicated researchers from Cardiff University are researching genetic aspects of FH - Dr Pauline Ashfield-Watt (part-time, pictured left) and Ms Kate Haralambos (full-time, pictured right). You may already be familiar with Pauline and Kate - they have both worked in the FH office at Cardiff University in various roles. Kate developed the patient management system for the FH service and Pauline has been involved in producing the FH Family Forum Newsletter and organising the FH Family Walks.

Ongoing research -Wales

The All Wales FH Cascade Testing Service has been a major step forward for patients with inherited high cholesterol. Genetic testing for alterations in genes (spelling mistakes in the genetic code) that cause FH is an effective way of finding family members who are at risk of heart disease before they have clinical features of FH. To date, we have identified genetic spelling mistakes in 25% of the patients that we have tested. This means that for 75% of people tested we are not able to provide a definite genetic diagnosis. This may be because there are other genetic spelling mistakes that we don't yet know about or don't yet test for. The FH research team are working with the FH clinical team (doctors and nurses) to remedy this. Two new studies are underway which aim to improve things.

Variants of Uncertain Significance Study (VUS Study)

Sometimes we identify a genetic variant in one of the three main FH genes but cannot be sure if this is actually causing FH or not. These are called variants of uncertain significance (VUS). A VUS is found in around 8% of index patients. An index patient is the first person in a family who is diagnosed with FH. Currently, family genetic testing for VUS is not routinely offered because we don't know if the VUS is having an effect on cholesterol levels or not. Kate is coordinating a project for the families of index patients with a VUS to try and find out if a VUS causes high cholesterol. This project is led overall by Dr Ian McDowell with the support of all Lipid consultants and FH nurses in Wales. Kate's work on this project is supported by the Heart Research Fund for Wales.

Who can take part in the VUS study?

Kate will be working with the Clinical Care Team to contact index patients with VUS to offer genetic testing to their families and to assess whether the VUS tracks with cholesterol levels in the family. This will help to determine whether the VUS is disease causing and improve the genetic diagnosis and treatment of FH. You can read more about this study on the FH Wales website **www.fhservice.wales.nhs.uk/research-and-development**

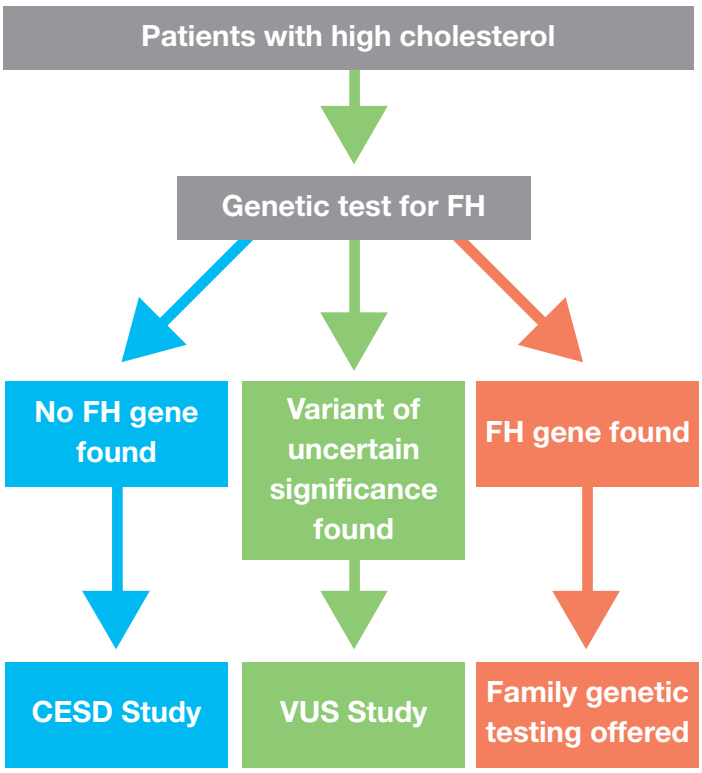
CESD Study - Are there other genetic spelling mistakes that cause high cholesterol?

About two thirds of the index patients that we test for FH do not have any alteration detectable in the three main FH genes. This does not mean that they don't have a genetic cause of high cholesterol. It just means that the genetic cause or causes of their raised cholesterol levels haven't yet been found. DNA samples from all patients that participate in the FH testing service are stored so that they can be re-tested if new genetic tests become available. Recently, a condition called Cholesteryl Ester Storage Disorder (CESD) has been reported to resemble FH clinically and is linked to premature coronary artery disease. CESD results from a genetic alteration. We are about to start a project that will use stored samples from FH index patients with "negative genetic results" to find out how common the CESD alteration is in patients with inherited high cholesterol, but who do not have one of the common FH genes.

Who can take part in the CESD Study?

If you were the first person in your family to have an FH genetic test (index patient) and you had a negative result then you can be tested for the CESD gene alteration. The research team will need your consent to test your stored DNA sample. You will soon receive a letter from the Clinical Care Team telling you about the study.

You don't have to wait for this though – if you want your sample to be included in the study please contact Pauline on 02920 743864 or email ashfield-wattp1@cardiff.ac.uk to request a study information sheet and to enrol on the study. You can also download these forms from our website and post them to us directly.
www.fhservice.wales.nhs.uk/research-and-development



Other News

Living with FH

An FH patient from Ceredigion shares her family's FH story in an interview with the British Heart Foundation in an article from the May/June edition of 'Heart Matters' **www.bhf.org.uk/heart-matters-online/may-june 2014/medical/focus-on-fh.aspx**

Double honours for FH Specialist Nurse

The work of FH Specialist nurse Rob Gingell has recently been acknowledged by two charitable committees. At the recent British Cardiac Society/BHF awards ceremony it was announced that Rob is their 'Highly Commended 2014 BHF Ambassador'. He has also been awarded the Rianna Wingett Award 2014 by Heart UK. Rob was nominated for the award by an FH patient who praised Rob's work in North Wales and commented "I believe that Rob absolutely cares about FH and his future patients are lucky to have him fighting their corner." Congratulations Rob, keep up the good work.

Please help us to stay in contact with you.

Approximately 1600 of these newsletters are being sent out to index patients who have been tested for the FH gene and their relatives who have had a positive genetic test. Of these, only 102 patients will receive the newsletter by email directly from the FH Family Forum. This special edition of the newsletter has been sent to you via the FH Wales Service and funded by FH Research.
We cannot guarantee that future editions of the newsletter will be supported in this way. The FH Wales Service cannot provide your contact details to the FH Family Forum because of Data Protection Policy. This means that unless you provide your contact details to the FH Family Forum we will not be able to contact you directly. If you want to receive information from the FH Family Forum, including future newsletters please provide your contact details listed below by email or post to FH Family Forum, C/o FH Wales Service, Medical Genetics, SAC Building, University Hospital of Wales, Cardiff, CF14 4XW. Thank-you, we really look forward to hearing from you.

FH WALES **FH Family Forum**

Your contact details

Title

First name

Surname

Email

Address

Postcode

Tel (home)

Mobile

Email: fhwales@gmail.com

Phone us via the FH Service office:
FH Office: **029 20744021**

Website:
www.FHwales.co.uk

Facebook:
FH for short

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