# All Wales FH Family Forum

-for individuals and families with inherited high cholesterol

FH WALES
Familial Hypercholesterolaemia

Spring 2017

## FH Testing Services

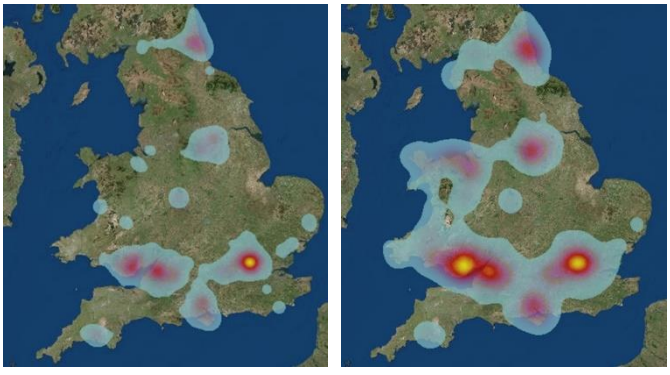
### 1000 Genetic FH Diagnoses in Wales

We have now identified 1000 people with a genetic diagnosis of FH in Wales which is great news. Recent studies suggest that FH is more common than previously thought (approx. 1 in 250 people in the UK), so we still have plenty of work to do finding and treating people with FH.

### FH services spread across the UK

Since 2010 Wales has been leading the way with a national FH service. More English regions are now on board too with 27 FH nurses appointed across 12 sites in England with support from British Heart Foundation. Northern Ireland and Scotland also have established FH services.

The 'heat maps' below show the spread of FH genetic testing across England and Wales between 2012 (left) and 2017 (right).



We are supporting these new services by sharing our experience. Kate Haralambos is helping to train nurses to use the FH Register and Cascade Testing Management System "PASS" that was developed for the Welsh service.

### Changes to the FH Clinical Team

We bid farewell to Rhiannon Edwards (FH Clinical Nurse Specialist) who has joined the British Heart Foundation as a Clinical Development Co-ordinator. We are very grateful to Rhiannon for her contribution to developing FH services particularly nurse led FH testing clinics in South East Wales and to the FH e-learning package. She will be able to link in with the FH service in her new role.

From May 2017, the FH Clinical Nurse Specialist for the South East will be Amanda Prosser. In South West Wales David Simpson with be joining Delyth Townsend, who has reduced her hours to part time.

The situation in the North is unchanged where Rob Gingell is the Clinical Nurse Specialist.

Mark Hale is the FH Paediatric Nurse.

### PCSK9 Inhibitors – A new cholesterol lowering treatment

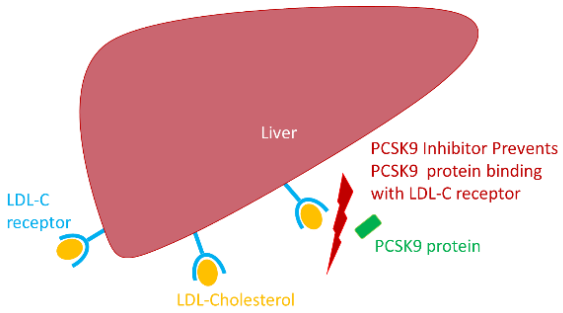
Standard medications (statins and ezetimibe) work well for many people with FH, reducing cholesterol and heart attacks. These drugs are very safe and most people get on well with them. Some

people, however, do have adverse effects such as muscle aching (about 5%) and for others cholesterol remains high despite maximum treatment.

PCSK9 inhibitors are a new treatment that has recently been approved for use in Wales by the National Institute for Health and Care Excellence (NICE). Currently, alirocumab (Praluent) and evolocumab (Repatha) are available.

### How do they work and are they safe?

PCSK9 is a protein that circulates in the blood, links with receptors for Low Density Lipoprotein Cholesterol (LDL-C) on the surface of the liver and destroys them. LDL-C receptors are important for removing LDL-C from the blood. PCSK9 inhibitors work by preventing the PCSK9 proteins from destroying LDL-C receptors so that more LDL-C is removed from the blood.




The PCSK9 inhibitors need to be given as an injection every 2 or 4 weeks and are only available for use under supervision of a Lipid Clinic consultant.

These drugs are still very new, but studies show that they lower LDL-C well and appear to be safe and tolerated even by those individuals who have side effects from other treatment.

### Who is suitable for PCSK9 inhibitors?

FH patients who have persistently high LDL-C despite taking the maximum dose of standard treatments may be eligible. For people with FH who have cardiovascular disease, treatment may be considered if LDL-C is greater than 3.5 mmol/L and, for those without cardiovascular disease, if LDL-C is greater than 5 mmol/L.

## Service Development

### A New Web Tool to Help Select Patients for FH Genetic Testing

It is important to offer FH genetic testing to those patients who are most likely to have an FH gene. In Wales a set of five criteria are used to provide a score that helps the clinical team to estimate the likelihood of a patient having a positive genetic test for FH. This forms the basis of the discussion with patients about whether they wish to be tested. Recently the research team has developed this scoring system into a web tool. We hope that the web tool will be used in clinics across Wales during 2017 and then rolled out across the UK.

## Research update

### Variants of Uncertain Significance Study (VUS Study)

Sometimes when we do genetic testing we find genetic variants, but we don't know whether they are causing FH or not. This study is helping to identify genetic variants which cause FH. Studies on 14 genetic variants have now been completed. Nine were found to cause FH, four did not and 1 remained unknown. The study is ongoing.

## Cholesterol Ester Storage Disorder Study (CESD Study)

Thank you to all the patients who consented for their stored DNA samples to be re-tested in this study. CESD is a rare genetic cause of high lipids. It differs from FH in that it is inherited as a genetic recessive condition (does not show if you have only one copy of the gene) unlike FH which is genetically dominant (a single copy of the gene causes high cholesterol). In other countries, some patients with CESD have raised LDL-C levels that resemble those found in FH. This led us to think that some patients who had previously tested negative for the standard FH genes may have CESD.

In total 663 patient DNA samples were analysed. Of these, three patients had a single copy of the CESD genetic variant (mutation) but no-one had two copies of the CESD mutation. This means that we did not find any patients with Cholesterol Ester Storage Disorder. For the three patients who carry a single copy of the CESD genetic variant, this was not of any clinical significance, because of the recessive nature of the gene.

The frequency of the CESD variant in our Welsh FH patient group (1 in 221 patients) is similar to that in other parts of Europe. This study is now complete and was presented at the 2016 HEART UK Conference in Edinburgh. We continue to work with the laboratory to find new genetic causes of FH.

# Young people's section

## Involving FH Families.

The first National FH Family Day was held by HEART UK last October. The event at Missenden Abbey was attended by families from across England and is planned as a forerunner to Regional Family Days which will be held across the UK during 2017. It was a fun and educational day. Talks were given by FH experts, and families were able to ask questions and meet other families with FH. Children and young people took part in discussion groups and were entertained by a magician. Dr Pauline Ashfield-Watt from the Wales FH Research Team talked about her forthcoming film making project with children and young people in Wales. Details of the Family Day can be found online at

https://heartuk.org.uk/annual-ambassador-family-day/national_fh_family_day

## Children and Young People's Videos

Pauline Ashfield-Watt and videographer Alex Philips have been working with children with FH to create a fun experience where children can learn about the condition, share their experiences and put together short videos about living with FH. The first workshop was held on Saturday 1st April 2017 and five young people learned about sketch noting, storyboarding and brainstormed key questions that could help newly diagnosed FH patients to live with FH. At the next workshop they will


record video footage and audio voiceover and edit these into one or more short videos to be shared on the FH Wales and

HEART UK websites. The project is supported by HEART UK and Cardiff University.

## Other news

### FH Wales to the fore in Westminster FH lobby

On 22nd March representatives from the FH Family Forum, the FH Wales Service, HEART UK and the British Heart Foundation attended an event in Westminster hosted by the All Party


Parliamentary Group on Heart Disease. Suzanne Sheppard who attended on behalf of the FH Family Forum had this to say about the event,

*"Attending the event at Westminster was quite an experience for my son and I. It was good to speak to the MPs and encouraging that they took an interest in our condition and the importance of finding FH families early. Moreover, it was really interesting to speak to other FH patients and nurses from around the UK to compare how their service works and to find out how other patients are treating their condition."*

## Please help us to stay in contact with you.

We do not have funding to send out these newsletters regularly by post. Please submit your email address to the FH Family Forum to receive these newsletters electronically in the future. To join the FH Family Forum and participate in activities to support families with FH please email us or complete and post the form below.

### Contact details for the FH Family Forum

**Email:** **fhwales@gmail.com**

**Phone:** **029 20744021 (FH Service)**

**Website:** **www.FHwales.co.uk**

**Facebook:** **FH for short**

**Post:** **FH Wales Service, Medical Genetics, SAC Building, UHW, Cardiff, CF14 4XW.**

*Please note: email addresses will not be used for discussing individual patient care.*

### FH Family Forum membership

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