



# FAMILIAL HYPERCHOLESTEROLAEMIA PAEDIATRIC REGISTER NEWSLETTER

Website: <https://audit.rcplondon.ac.uk/PaedFH/page.aspx?pc=homepage>

Dear Colleagues

Welcome to the first register newsletter which we plan to send out 2-3 times per year. We are all very excited that the project has finally got off the ground, with REC approval thanks to the hard work by Nancy Turnbull and Uma Ramaswami on getting the answers clear and accurate, so we only needed some minor changes to get the full approval. Also a big thanks to all the clinicians who have enrolled and are ready to start registering their patients - we are keen to help you in any way we can and are asking you to fill in a questionnaire about this to identify and remove as many blocks to registration as we can. Finally, we hope to see you at HEARTUK in Bristol in July, or to come and visit your centre if that would be helpful.

Best Wishes

Steve Humphries Chair of Steering Group

### ***ETHICS COMMITTEE RESULTS***

The register received Research Ethics Committee Approval on 30th January for a period of five years REC reference 12/NE/0398.

The [full application](#) and [favourable opinion letter](#) can be downloaded from the website for information. The conditions of research governance, is detailed in the favourable opinion letter. If you have questions or would like further information, ***please contact the Project Manager at the email address below.***

### ***FREE DNA TESTING FOR REGISTERED CHILDREN***

We are pleased to offer to clinicians entering data to the FH Children's Register, the possibility of sending blood samples for genetic testing to the British Heart Foundation funded Cardiovascular Genetics Research Laboratory at UCL. The laboratory, which is headed by Professor Steve Humphries, has a great deal of experience in screening for FH-causing mutations (1) and interpreting the potential pathogenicity of novel mutations (2) and are happy to provide this for any registered child with a family mutation that has not been identified. Currently the methods used are a high resolution melting technique to screen all coding

### ***27,967 PATIENTS LEFT TO REGISTER!***

There are estimated to be about 28,000 children and young people under the age of 18 with FH in the UK and the aim is to collect long term information on all of them. To date we have 33 children registered from 41 clinicians who have enrolled.

Please pass this newsletter on to colleagues who see children with FH.

### ***WHAT'S STOPPING YOU?***

We would like to understand what you find are the barriers to recruiting patients and entering their data on the database so that we may work with you to overcome them. Please complete a [very short](#) questionnaire which is attached with this newsletter and return it to the Project Manager at the address below.

### ***CONSENTING PATIENTS***

Please obtain assent of children under 18 as well as parent/guardian consent. If a child is over 16 or a minor of any age who is 'Gillick competent' and does not assent to the treatment, please do not

***The FH Paediatric Register receives funding from Royal College of Physicians Trust Funds, the British HeaRT***

FH Newsletter Volume 1<sup>st</sup> March 2013

This letter has been sent to all the healthcare professionals who have enrolled to register patients. Please pass it on to colleagues. Contact Details: Nancy Turnbull, Project Manager [fh@rcplondon.ac.uk](mailto:fh@rcplondon.ac.uk)

exons and the promoter of the LDLR, as well as specific regions in the APOB and PCSK9 gene where the common FH causing mutations identified in the UK occur (1). In the future, next generation sequencing on library capture of all FH causing genes will be used (3).

Individuals who would like to send samples should contact the Project Manager at the email address below to receive a pro-forma and DNA consent form and instructions on how to take and send the sample. The sample will be given a unique ID and included as part of the on-going research activities and we generally hope to return a diagnostic report within three months from receiving the sample. It should be noted that CVG is a research laboratory and not an accredited diagnostic laboratory, and therefore for use in future cascade testing it is recommended that carriage of the identified mutation be confirmed in an accredited diagnostic laboratory.

1. Whittall RA, Scartezini M, Li KaWah, Hubbart C, Reiner Z, Abraha A, Neil HAW, Dedoussis G, Humphries SE. Development of a high resolution melting method for mutation detection in familial hypercholesterolaemia patients. *Ann Clin Biochem.* 2010; 47: 44-55.
2. Usifo EL, Leigh SE, Whittall R, Lench N, Taylor A, Yeates C, Orenge CA, Martin, ACR, Humphries SE Low Density Lipoprotein Receptor Gene Familial Hypercholesterolemia Variant Database: update and pathological assessment. *Annals of Human Genetics.* *Ann Hum Genet.* 2012 Sep;76(5):387-401
3. Futema M, Plagnol V, Whittall RA, Neil HAW, Humphries SE on behalf of the Simon Broome Register Group and Hurler M on behalf of the UK10K Consortium. Use of Agilent exome capture assay and Illumina deep sequencing technology as a diagnostic tool for Familial Hypercholesterolaemia. *J Med Genet.* 2012 Oct;49(10):644-9

### ***FH PAEDIATRIC REGISTER STEERING GROUP***

The Steering Group meets 2-3 times a year and last met on 1st February. Dr Peter Dale of Aneurin Bevan Health Board – and Cascade testing lead was welcomed to the group. More information on the Steering Group, including members is listed on the website.

[https://audit.rcplondon.ac.uk/PaedFH/page.aspx?pc=s\\_group](https://audit.rcplondon.ac.uk/PaedFH/page.aspx?pc=s_group)

enrol them as their wishes should override the consent of the parent/guardian. Please, if appropriate, consider asking for consent/assent again in a year's time.

All of the consent and assent forms and further information on consent are available on the website under the menu 'Information and Consent' on the homepage <https://audit.rcplondon.ac.uk/PaedFH/page.aspx?pc=homepage>

### ***CONFERENCES AND SEMINARS***

- We will have a stand at the HEART UK conference in Bristol July 3-5<sup>th</sup> 2013
- Prof Humphries and Dr Ramaswami are available to attend local or regional meetings to discuss the clinical management of children with FH and the importance of the Register to research in this field.

***Please contact the Project Manager at the email address for more information***

### ***CLINICAL AUDIT***

Most of you expressed interest in sharing information for clinical audit purposes. Here is some information that may be of interest when we have recruited more children ....

- Median (range) age when statins are started and whether that differs by gender.
- Diagnosis
- % on statins
- % on other therapies

***Please contact the Project Manager at the email address below let her know what you would be interested in comparing.***

### ***NEW RESOURCES***

Information and resources have been added to the website including:

- data collection proformas to collect details for initial registration and follow-up.
- guidance on consent and assent
- all of the consent and assent information and forms under the menu 'Information and Consent' on the home page.

***The FH Paediatric Register receives funding from Royal College of Physicians Trust Funds, the British Heart***

FH Newsletter Volume 1<sup>st</sup> March 2013

This letter has been sent to all the healthcare professionals who have enrolled to register patients. Please pass it on to colleagues. Contact Details: Nancy Turnbull, Project Manager [fh@rcplondon.ac.uk](mailto:fh@rcplondon.ac.uk)