

NEWSLETTER

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FH Europe - The European FH Patient Network

FH Studies Collaboration

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FHSC Closed Steering Meeting



FHSC closed annual meeting, consistently rated highly by committee members, was attended by 58 invitees plus more joined online.



FHSC Steering post-lunch facilitated re-gathering of colleagues, counterparts, collaborators, and friends after a couple of years of absence.

FHSC Coordinating Centre, Imperial College London welcomes new Research Fellow Dr Amany Elshorbagy

(Bottom photo, left – right: Dr A Elshorbagy, Dr AJ Vallejo-Vaz, Dr ARM Lyons, KI Dharmayat, Prof KK Ray, Dr J Brandts, B Cerutti)



The latest global collaborative work of the FHSC was represented at the EAS 90th Congress, May 2022, Milan Italy, via two well-received talks:

- Kanika Dharmayat *et al.* on behalf of EAS FHSC Investigators. Global perspective of paediatric Familial Hypercholesterolaemia: Analysis from the EAS FHSC Registry on over 11,200 children and adolescents with Heterozygous Familial Hypercholesterolaemia from 44 countries. Late Breaker at EAS 90th Congress 2022, Milan Italy.

https://www.youtube.com/watch?v=ycEyDTTD8_U

- Antonio J. Vallejo-Vaz *et al.* on behalf of EAS FHSC Investigators. Identification, characteristics and management of adults with heterozygous familial hypercholesterolaemia in high and non-high income countries participating in the EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). Science at Glance at EAS 90th Congress 2022, Milan Italy.



FHSC booth in the Exhibition room hosted by the Coordinating Centre Investigators was well-attended by our global counterparts and collaborators, supporters from industry, and representatives from the European FH Patient Network (FH Europe).

(Left – right: FHSC Chief Scientist Dr. AJ Vallejo-Vas, FHSC Data Manager and Software Engineer Mr CA Stevens, FHSC Research Associate Project Manager Dr. ARM Lyons, and FH Europe CEO Ms Daccord and colleague)

The EAS FHSC now spans 69 countries (shaded map) and includes 82 Lead Investigators; specifically the National Lead Investigators are listed [here](#). EAS FHSC Registry includes approximately 70,000 cases across 66 countries.



Become part of the expanding EAS FHSC

Do you have an interest in FH, collect clinical and/or genetic FH data and are keen to contribute to the **EAS FHSC Global Registry** ([CT.gov Identifier: NCT04272697](#))?

If so, we would like to hear from you!
For enquires contact info@eas-fhsc.org

More information about the EAS FHSC can be found in these publications:

- [FHSC Study Protocol: 'Pooling and expanding registries of FH'](#)
- [FHSC Survey: 'Overview of the current status of FH care in over 60 countries'](#)
- [FHSC Results: 'A global perspective on FH: Cross-sectional study from the EAS FHSC'](#)

FHSC Coordinating Centre provides a free essential web-based resource exclusive to FHSC Investigators and their local teams to support entering and managing local-level data, and sharing data with the FHSC Global Registry. Ask the Coordinating Centre for more details.

**Contribute to the
Worldwide Directory of Lipid Clinics & Patient Support Groups:
findmylipidclinic.com**

Follow link to register your lipid clinic and/or patient support group

Update on Portuguese FH Study

The Portuguese FH Study, coordinated by National Institute of Health since 1999, aims to identify and characterise genetic causes of hypercholesterolaemia in individuals that meet FH clinical criteria to improve prognosis by a personalised medicine approach. This approach includes improved diagnostic methods to characterise the FH phenotype and functional characterisation of variants causing FH to inform treatment.

Between 1999 – 2021 over 50 centres countrywide (distribution shown in Figure 1) referred 3,188 cases to our lab. Of these, 1,107 were index cases that met FH clinical criteria, and 2,081 were relatives. We identified the cause of FH in 396 of these index cases, being 10 homozygotes, and further cascade screening via genetic diagnosis identified 930 relatives with FH (318 children [mean age 10 years] and 612 adults [mean age 43 years]).



Figure 1.

Most of our patients have a pathogenic or likely pathogenic variant in the LDLR (92%) and only a small part has FH due to three different APOB variants (5%) and only 1% have a pathogenic variant in PCSK9. Interestingly, 2% of the genetically characterised patients have others monogenic causes for their hypercholesterolemia (3 with lysosomal acid lipase deficiency, 3 with dysbetalipoproteinemia, 1 with sitosterolaemia, 1 with analbuminemia) (Figure 2).

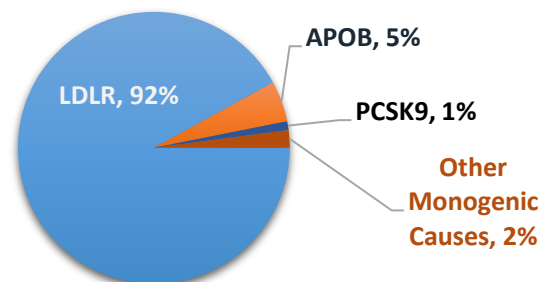


Figure 2. Genetic causes of FH phenotype

More than 140 different variants in the LDLR have been found in Portugal, being 8 variants responsible for 45% of all FH cases (Figure 3). Almost all missense and splicing Portuguese variants (~90%) have been functionally characterized, mainly by our lab.

24% of index cases with FH had already suffered a premature cardiovascular event (men <55 years, women <65 years), with a mean age 1st event of 45 years, as well as 9% of the relatives, with a mean age of 1st event of 39 years.

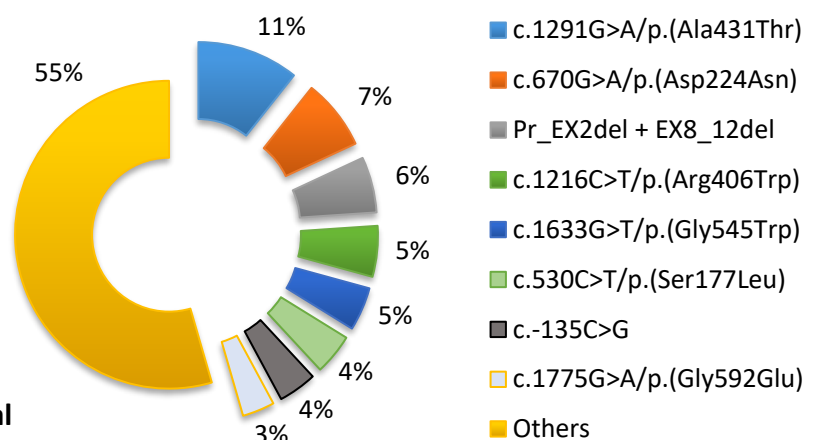
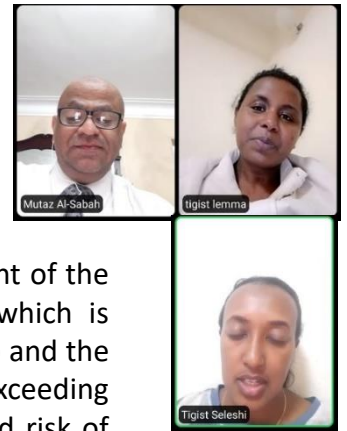


Figure 3. Most common variants in LDLR gene in Portugal.

Prof. Mafalda Bourbon
National Lead Investigator of Portugal

First FH Symposium in Ethiopia

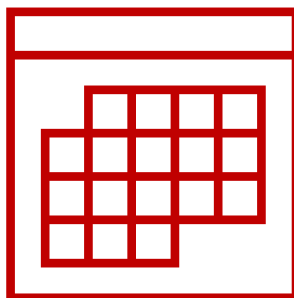
As part of our international advocacy efforts in awareness of FH in the African horn countries, and in collaboration with our colleagues in the Faculty of Health Sciences, Addis Ababa University in Ethiopia (Assistant Professor Tigist Seleshi and Assistant Professor Tigist Lemma), we arranged the first FH symposium there which was attended by a large group of physicians in the City.



This is the first step before starting the bigger project there (establishment of the first lipid clinic in that country). Ethiopia was one of the countries which is suffering from great challenges (Economic, health, and political instability) and the medical community there are in areas of unmet needs for a population exceeding 115 million. The FH symposium included information on (Prevalence and risk of FH, who to diagnose and treat, and the importance of early detection).

Dr. Mutaz Alkhnifsawi
National Lead Investigator of Iraq

**NEXT 2022 QUARTELY NEWSLETTERS
WILL BE ISSUED IN
SEPTEMBER & DECEMBER**



A unique, global, research launched on International Day of Action for Women's Health - 28th May

On the occasion of the International Day of Action for Women's Health, FH Europe partnered with [Erasmus University Medical Center](#), [Stichting LEEFH](#), University of Oslo, [Department of Nutrition](#), National Advisory Unit on FH ([NKTforFH](#)), [Oslo University Hospital](#), to launch an international survey on women and FH/HoFH. The aim is to better understand how the inherited high cholesterol affects women, their family planning decisions and health pre-, during and post- pregnancy. The goal is to engage 1000 women globally, in their local language. Would you be interested in promoting the survey among your female patients to advance the knowledge and improve the standard of care?



If so, visit [FH Europe's website](#)

FH Paediatric screening – moving prevention from evidence to action. Overcoming the barriers to implementation.

Since accepting Paediatric FH screening European Commission to the Public Health Best Practice Portal, FH Europe together with its Network and partners, has been advocating for FH paediatric screening across Europe.

In October 2021, FH Europe held a high-level Technical Meeting under the Slovenian EU Presidency which generated a strong consensus, outlined in a call to action on FH screening published in the European Heart Journal.



Yet still paediatric FH screening programmes are rarely implemented due to a blend of barriers (finance, health system, public lack of trust or awareness, lack of political leadership to champion screening). Join in person or online the high level meeting under the auspice of the Czech EU Presidency prepared in partnership with Diagnostica FH. More on [FH Europe's website](#).

Magda Daccord, FH Europe Chief Executive