EAS FHSC NEWSLETTER

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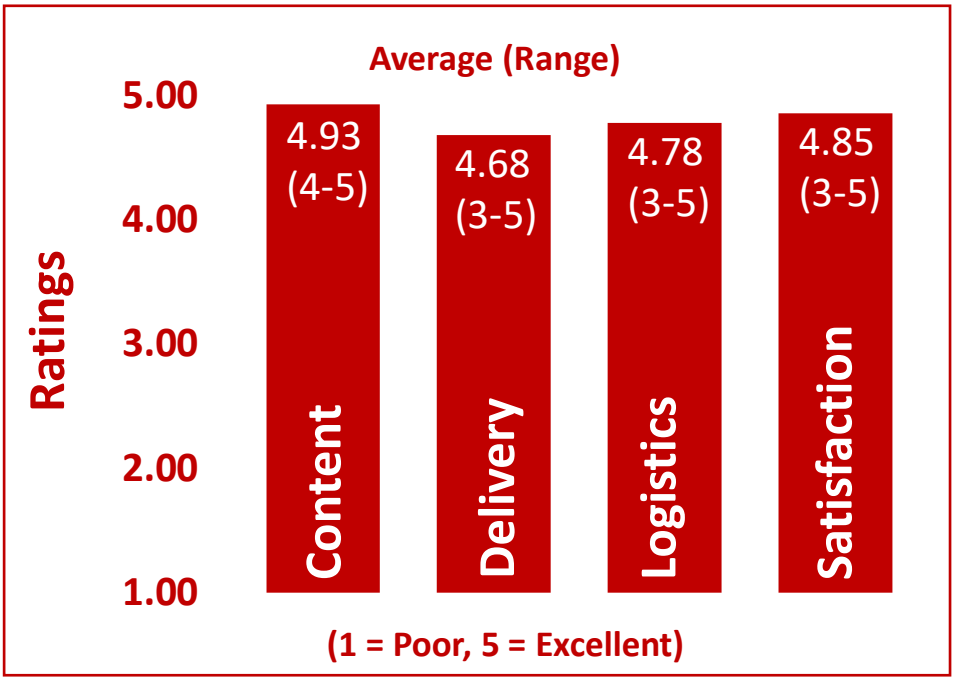
Steering Committee Meeting 2021 Report

This Annual Closed Meeting held in conjunction with the 89th EAS Congress 2021 included 73 participants.

The Agenda included a formal briefing from the Coordinating Centre followed by a series of guest speakers comprising National Lead Investigators (see page three for their reports) and representation from the European Patient Support Group 'FH Europe' (see next page for their report and latest news).

This Meeting was well received and the feedback ratings by the participants are shown in the figure below.



Next Steering Committee Closed Meeting will be held on the morning of Sunday 22nd May

in conjunction with the

90th EAS Congress 2022, Milano Convention Centre (MICO), Milan, Italy
Sunday 22nd May - Wednesday 25th May 2022

FH Europe joins forces with European health organizations in a Call for EU Plan on CVD

On June 16th, FH Europe together with 11 other European health organizations signed a **joint statement** calling on the EU to develop a comprehensive plan regarding CVD prevention and treatment.

Although cardiovascular mortality has declined over the past 50 years, thanks to investment in public health, prevention, and acute care, CVD remains the leading cause of death in the EU. It accounts for 36% of all deaths and around 20% of all premature deaths. The COVID-19 pandemic has worsened this scenario, causing additional damage to the hearts and vascular systems of European citizens. Policymakers must act now to prevent CVD from becoming the next pandemic.

The signatory organisations jointly call on EU decision-makers to develop a comprehensive EU Plan on CVD to ensure that citizens can live longer, healthier lives – regardless of where they are born or live in the EU – and enable them to continue to contribute to society and the economy.

FH Europe's Chief Executive, Magdalena Daccord says "Our aim is to bring to the attention of the EU policy makers the voice and the needs of our international patient network, their families, and the European citizen. These individuals are impacted by inherited, genetic lipid conditions, primarily FH and elevated lipoprotein (a), which are non-modifiable. Through this collaboration, we will act on the recommendations from the Global Call to Action on FH compiled by the global FH community, including EAS and EAS FHSC. We want to specifically lobby for childhood screening for FH, based on the FH paediatric screening best practice, accepted by the European Commission Public Health Best Practice Portal".

"Innovation - data, genomics, algorithms - will transform the traditional image of CVD in the years to come. For most cardiovascular diseases, prevention will become increasingly personalised (compared to the current model "one size fits all"), screening and early detection of diseases will appeal to genetic characteristics and genomic data. In the heterogeneous and complex picture of CVD, FH is already a model in which innovation has entered and could be used as a perfect example. To show value to all European patients and citizens, however, we need collaboration, a new vision, and a unitary implementation of CVD innovations at the level of the European Union" added Marius Geanta, who together with Samuel Gidding co-chairs FH Europe's Scientific and Public Health Advisory Committee.

Magdalena Daccord,
FH Europe Chief Executive

Steering Committee Meeting Guest Speakers' Reports

Achievements in FH Awareness and Management in Iraq

Despite challenges of poor economy and 1/3 of Iraq being occupied by terrorist groups, our team achieved great steps in awareness and management of FH in Iraq:

- Establishment of 12 lipid clinics across Iraq, providing healthcare to FH patients, visit www.iraqilcn.com.
- Launch of Electronic FH Consultation Centre, an e-system of communication between patients and physicians during the pandemic, visit www.fhe-center.jmu.edu.iq.
- Establishment of our Patients' Advocacy Group, lead by a female HoFH patient, to raise patient voices to participate in decision making, contact iraqifhadvocacy@yahoo.com.
- Our national paediatric FH screening runs in nine paediatric lipid clinics. Also given the consanguinity in Iraq, we launched our unique optional support service to couples planning to start their families via premarital FH screening.

We thank the International Atherosclerosis society (IAS) and Oman Society for Lipidology and Atherosclerosis (OSLA) for supporting our project, and to our colleagues in Al-Mamorah Scientific Bureau for sponsorship.

Dr. Mutaz Al-Khnifsawi, FHSC NLI of Iraq

HICC HoFH Global Registry

Ever since the launch of this initiative several years ago, we have steadily expanded the HICC Registry, making new contacts with many colleagues caring for patients with homozygous familial hypercholesterolemia (HoFH) across the globe. We are now proud to present the first fruits of our labour in the form of a manuscript ready to be submitted in a high impact journal, that reports on data from 750+ HOFH patients across 38 countries.

Data from our consortium allow us to paint a more precise picture of HoFH and its treatment; a striking observation from these analyses is the health disparities that exist between patients living in high-income countries versus the rest of the world. We believe that this has the potential to influence policy and eventually improve care for patients with this rare yet life-threatening disease globally.

We thank the members of HICC for their contributions and commitment to the care of HoFH.

Not yet part of HICC but interested? Feel free to reach out at: coordinator@eas-hicc.org

Prof. Frederick Raal
FHSC Executive & NLI of South Africa

Contribution by Dr. Tycho Tromp
HICC Coordinator

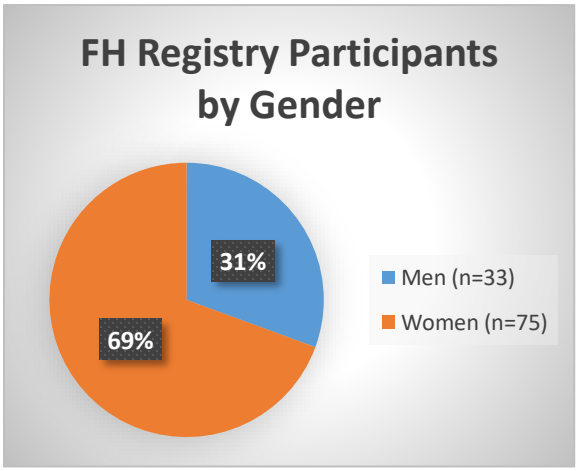
Disparities by gender among adults with HeFH in the Maltese FH Registry

The Maltese FH Registry was established in 2017 and has collected phenotypic data from the only Lipid Clinic and multidisciplinary Health Centres within the national health service since January 2017.

This year the data was presented at the 89th EAS 2021 Congress (poster ID 1270) aiming to describe FH characteristics by gender and identify areas for improvement from a cross-sectional analysis of entry data.

Overall 108 HeFH patients were included, with mean ages that were late for an inherited condition in the late 50s for women, which was on average 10 years later than in men (data analysis of Definite and Probable FH). Most were index cases, with few identified from cascade testing. Cardiovascular disease was more common in men and peripheral vascular disease in women. More men than women were on lipid lowering therapy at baseline. Median LDL-cholesterol values were similar by gender, both on and off treatment.

Overall, this analysis identifies that there is clearly a need for improved screening for earlier identification and preventive care and that gender disparities need consideration in the management of FH. (Data also contributed by C. Azzopardi, Lipid Clinic).



Dr. Myra Tilney, FHSC NLI of Malta

FH in India

FH in India is found to be more common than thought with an estimated frequency of 15% in CAD patients. Published FH data in India is scarce and limited to genetic studies with small sample sizes and sporadic case reports. To our knowledge there are only six genetic studies, and mutational spectrum for 37% FH cases is unknown. Of the three classical FH genes, the most studied are LDLR followed by ApoB.

Our recent article published in *Journal of Human Genetics*, Reddy *et al.*, 2021, screened the entire PCSK9 gene and reported a Loss-of-Function mutation (rs562556) for the first time in India along with seven other benign variants.

These six prior studies reflect genetic heterogeneity of Indian FH population as there are very few overlapping genetic variations found. Currently, our ongoing project funded by the Indian Council of Medical Research intends to perform whole exome sequencing on Indian FH subjects in an attempt to further understand the mutation spectrum.

Under aegis of the EAS FHSC, the India FH Registry have so far included data of more than 200 FH patients, with additional data to be included as more centres participate.

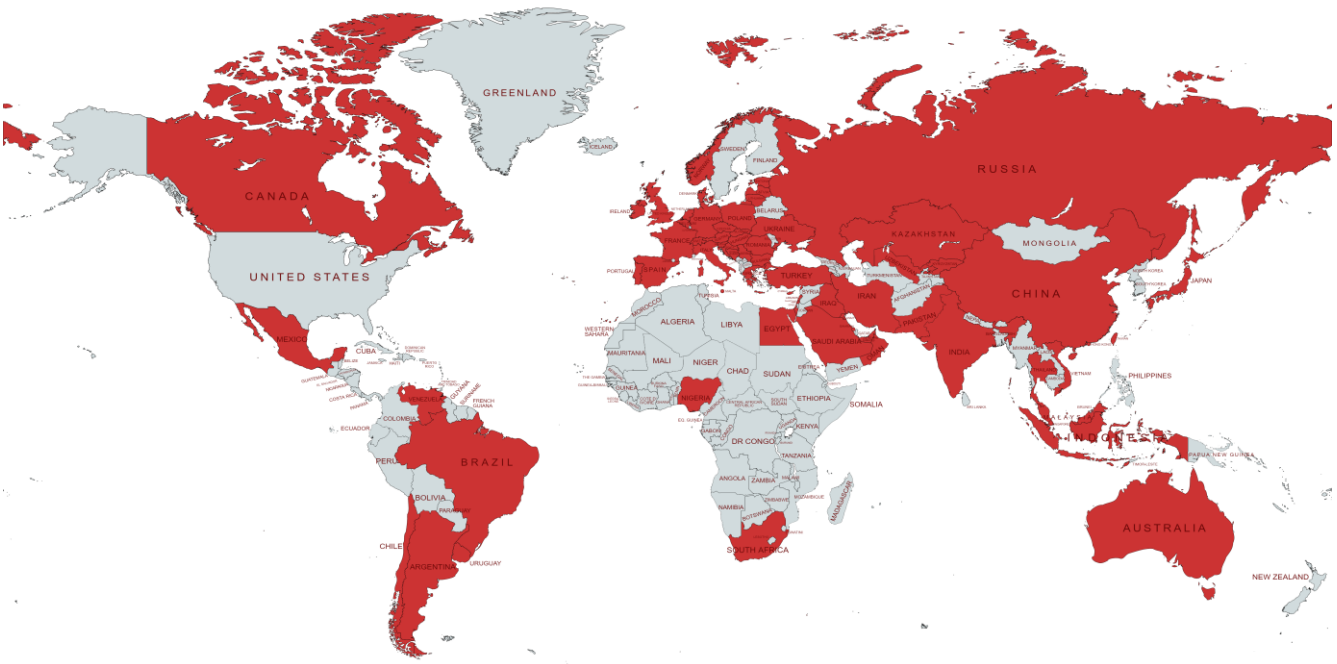
Furthermore, extensive nationwide screening programs should be encouraged to increase awareness and close the gap in detection, diagnosis and treatment for FH in India.

Dr. Tester F. Ashavaid, FHSC NLI of India

Dr TF Ashavaid (center), **Dr SAV. Shah**, Senior Molecular Scientist (Right), **Ms LL Reddy**, Senior Research Fellow

Global Network and Registry

The EAS FHSC spans 66 countries (see shaded red map below) and includes 80 Lead Investigators; specifically the National Lead Investigators are listed here. EAS FHSC Registry includes >66.4K cases across 65 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 open-access 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 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.

Interested to contribute an article (FH news, publications, events, etc) to the next EAS FHSC Newsletter?
Please contact: info@eas-fhsc.org || coordinator@eas-fhsc.org