



# FHSC

EAS Familial  
Hypercholesterolaemia  
Studies Collaboration

Summer 2024  
Issue 28

## NEWSLETTER

Enhancing Identification of Familial  
Hypercholesterolemia in Electronic Health  
Records with AI



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### Highlights from the Steering Committee Meeting



The recent Steering Committee Closed Meeting was held on May 26, 2024, in Lyon, France. Eighty people attended in person, while others participated online. The meeting featured several presentations, including updates from the FHSC Coordinating Centre on ongoing work and reports from the National leads. Below are some of the presentations and the speakers who participated in the event.

Below are some of the presentations:

1. FHSC Coordinating Centre
  - a. Dr Irene Karungi - Genetics Methods Survey
  - b. Dr Amany Elshorbagy - Genetics project classification - details on page 3.
  - c. Mr Christophe Stevens - Call for Follow up data and data management in the FHSC registry.
  - d. Mr Didac Llop Paredes - Association of Cardiometabolic Risk Factors with Cardiovascular Disease in Familial Hypercholesterolaemia Patients with Severe Elevation of LDL-C
2. National Lead Investigators and FH Europe Organization
  - a. Prof. Olivier Descamps and Dr Alexandra Sperone NLI Belgium – Strategies to collect follow up data.
  - b. Magdalene Daccord FH Europe Foundation – Promoting PERFECTO a project co-funded by the European Union. More details at [www.perfecto-fh.eu](http://www.perfecto-fh.eu)

We are excited to share the following two key points

1. Excellent progress has been made on classification of the Genetics data.
2. **The deadline to submit follow-up data is extended to 15 December 2024.**

We would like extend our appreciation to all FHSC National Lead Investigators for their dedication and efforts in responding to the call for genetics data.

**Please save the date for the next EAS – FHSC Steering Committee closed meeting, which will take place on May 4, 2025, in Glasgow.**

### Progress Made on the Ongoing Genetics Project



The FHSC Genetics project data collection and classification stages have been completed as of June 2024, with support from more than 40 FHSC National Lead Investigators and an expert FHSC Genetics Working Group. This is the largest database to date of the spectrum of DNA mutations and protein variants causing FH, as well as non-FH-causing variants discovered upon genetic testing for suspected FH.

The FHSC Genetics Project includes more than 40,000 patients with FH from more than 40 countries. The project aims to map the most common FH variants detected in various world regions and to provide practical recommendations for best practices in FH genetics testing. The project will also enhance our understanding of how genotype and environment, including geographical location, interact to determine disease severity and outcomes in patients with FH.

Interim findings from two of the project studies were presented at the 2024 EAS Congress in Lyon by the FHSC Researchers Amany Elshorbagy and Christophe Stevens.

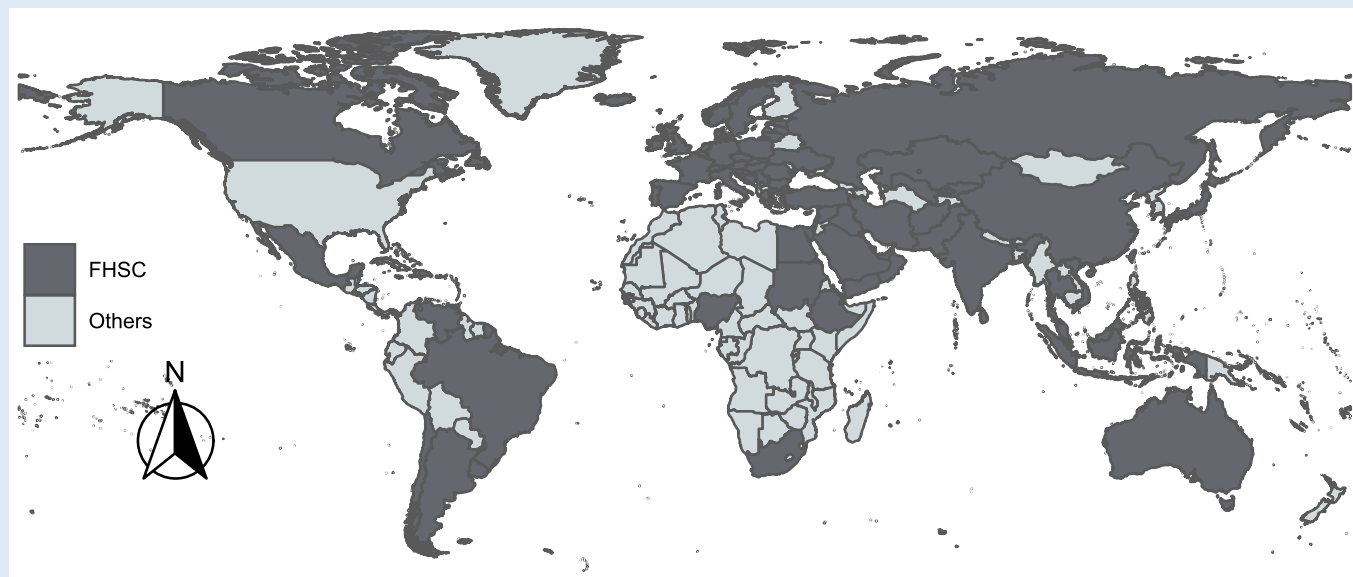


**Presentation 1: Spectrum of Gene Variants in Children and Adults with Heterozygous FH Across World Regions: A Study By The EAS FHSC**



**Presentation 2: To Correct or not to Correct: Estimating Pre-treatment LDL-C Concentrations in Genetically Characterized Patients with FH on Lipid-lowering Medication**

We express our gratitude to the FHSC Genetics Working Group for their ongoing support and commitment to the Genetics project.



**FHSC spans 76 countries and includes 92 [National Lead Investigators](#). The FHSC Registry includes approx. 80K cases across 68 countries.**

## GET INVOLVED

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](mailto: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'](#)
- [Overview of a collaborative global effort to address the burden of FH](#)
- [Familial hypercholesterolaemia in children and adolescents from 48 countries: a cross-sectional study - ScienceDirect](#)

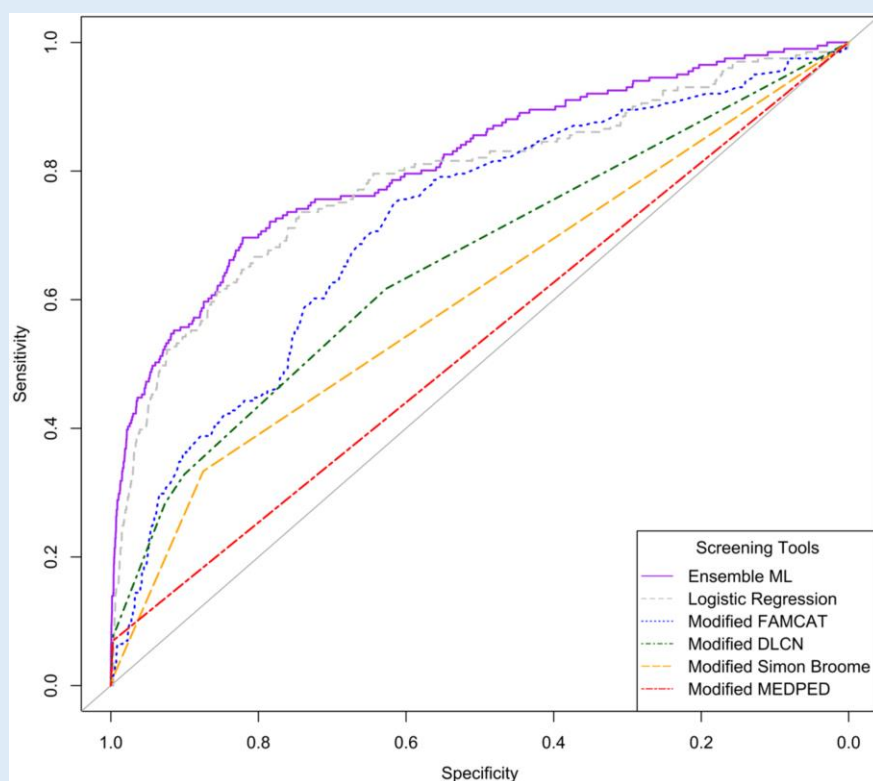
**FHSC Coordinating Centre** provides a free essential web-based resource exclusive to FHSC Investigators and their local teams to support entering and managing local-level data, and sharing data with the FHSC Global Registry. Ask the Coordinating Centre for more details.

**Worldwide Directory of Lipid Clinics & Patient Support Groups:** [findmylipidclinic.com](https://findmylipidclinic.com)  
For more information: [Stevens et al. EAJ 2022;2:37-40](#)



## Enhancing Identification of Familial Hypercholesterolemia in Electronic Health Records with AI

Researchers at the coordinating centre of the EAS FHSC have published a new article in the Journal of the American Heart Association (JAHA), showing that AI could help identify individuals carrying (likely) pathogenic variant(s) linked to FH in electronic health records (EHRs) with greater sensitivity and precision than existing clinical diagnostic criteria and the screening criteria recommended in the UK.



**Figure: ROC Curves of newly derived models and existing clinical diagnostic and screening criteria**

The best AI model derived in the study reduced the number of necessary tests by about one third to identify more carriers of FH variants compared to current screening methods. Implemented in EHRs, this tool could offer a scalable and potentially cost-effective solution to identify individuals most likely to receive a positive confirmatory genetic test for FH and ultimately help reduce the high number of cardiovascular events caused by this condition.



<https://www.ahajournals.org/doi/10.1161/JAHA.123.034434>

### Establishment of the First Ever Lp(a) Research Team in Iraq



A remarkable progress in the field of Preventive Cardiology services and care of FH patients in Iraq was achieved in collaboration with the health authority and medical education (Karbala, Iraq). In March 2024, we announced the launch of the first ever Lp(a) Research team in Khadeejat Al-Kubra Women's hospital after making this test available for the first time in Iraq In February 2024.

The entry of Lp(a) test and the establishment of the first ever Lp(a) Research Team there represents a quantum leap in Preventive Cardiology service and care of FH patients in Iraq.

We acknowledge the pivotal role of Dr. Haydar Al- abidi, head of health authority and medical education for making this great achievement possible and implementable .

**National Lead of Iraq ,  
Dr Mutaz Al-Khnifsawi**

## From Lyon to Budapest – building on patient advocacy in FH and elevated Lp(a) for Cardiovascular Health Plan for Europe



At the EAS annual Congress in Lyon, FHEF Patient Ambassadors played a prominent role, not only as speakers but also as exhibitors for the first time. They shared their experiences, the burden of disease, and their hopes for the future at the FH paediatric symposium, both in-person and via video in the main plenary session. Four international Patient Ambassadors, living with HeFH, HoFH, FCS, and elevated Lp(a), delivered powerful stories about their patient journeys. They discussed their diagnoses in childhood, daily challenges, experiences with clinical trials, psychological burden, and the latest advocacy efforts.

<https://fhf.org/news/eas2024/>

Simultaneously, the [Lp\(a\) International Taskforce](#), chaired by Prof. Florian Kronenberg, held an in-person meeting to discuss specific actions related to a new -

health economic analysis and the upcoming Global Lp(a) Summit in 2025.

In collaboration with the EAS, the FHEF also had a dedicated booth where Patient Ambassadors engaged with the medical community, eager to learn more about patient experiences, support, and the broader scope of FHEF. The booth was a focal point, especially being located opposite the Lp(a) testing station.

The advocacy work of FHEF Ambassadors, including patients, medical doctors, and researchers, has been widely recognized within the cardiovascular disease community. On July 4th, the FHEF delegation participated in [a high-level conference in Budapest](#), dedicated to the European Cardiovascular Health Plan under the Hungarian EU Presidency. Alongside health ministers from across the EU, they discussed familial lipid disorders (FH, elevated Lp(a)) within the context of inherited CVD risk factors. They highlighted the [Prague Declaration](#) and [PERFECTO FH](#) project, an EU co-funded initiative, as a best practice for FH paediatric screening, aiming to address CVD, the leading cause of death in Europe.

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**Interested to contribute an article (FH news, publications, events, etc)?**

**Please contact: [info@eas-fhsc.org](mailto:info@eas-fhsc.org)**

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