FHSC
EAS Familial
Hypercholesterolaemia
Studies Collaboration

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NEWSLETTER

CONTENTS

Your Data Matters: Extended Opportunity for FHSC Follow Up Data Submission

The FHSC Coordinating Centre extends its sincere gratitude to all National Lead Investigators (NLIs) and their teams for responding to our request and submitting follow-up data since 2023. Your efforts are invaluable in advancing the FHSC mission.

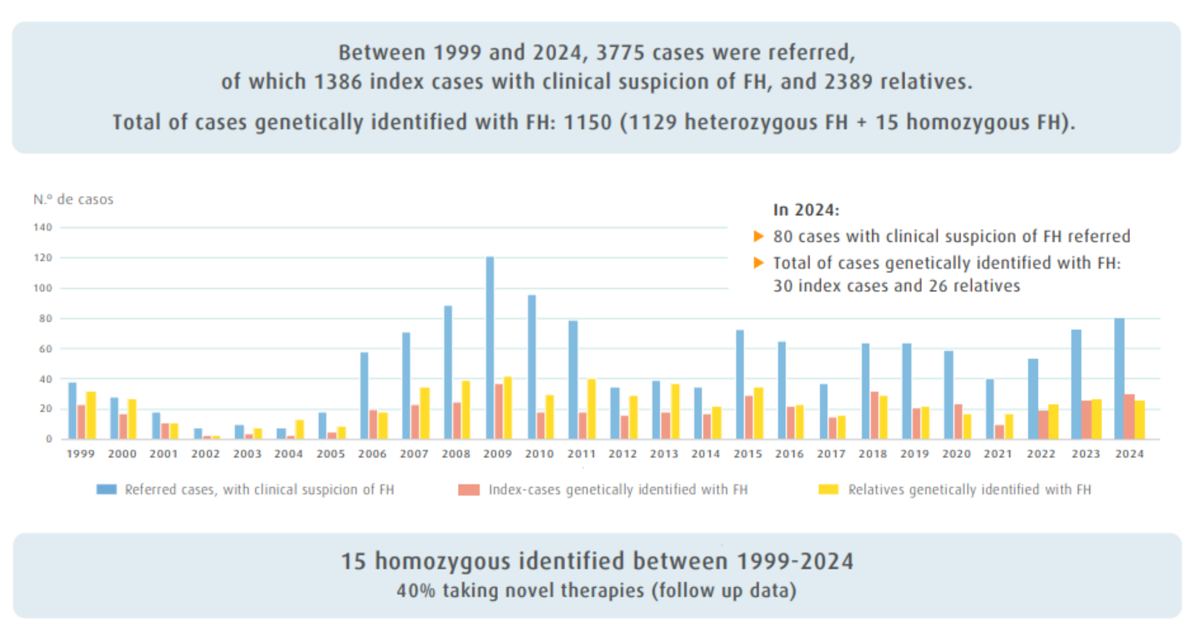
Although the official closing date of 31 October 2025 has passed, we are pleased to announce an extended opportunity for those who were unable to share their follow-up data. You can still submit these important datasets through the FHSC website. Additionally, teams with access to IDEAPP may continue entering patient follow-up information directly into the system.

Furthermore , we would like to inform all NLI's who have responded to the FHSC call for Follow- Up and have submitted their Follow – Up Data datasets that we shall be reaching out to you soon with data cleaning queries in areas where the data was incomplete, This process ensures that the analyses we conduct and the research findings we share are accurate and valid.

We deeply appreciate your collaborative support and the datasets submitted so far. Beyond follow-up data, we also encourage you to share any baseline and genetic data you may have, ensuring it is included in the FHSC Data Warehouse.

Thank you for your commitment to this global initiative!

Fighting FH Together: Progress and Impact Since 1999



Between 1999 and 2024, 3775 cases were referred, of which 1386 index cases with clinical suspicion of FH, and 2389 relatives.

Total of cases genetically identified with FH: 1150 (1129 heterozygous FH + 15 homozygous FH).

In 2024:
- 80 cases with clinical suspicion of FH referred
- Total of cases genetically identified with FH: 30 index cases and 26 relatives

15 homozygous identified between 1999-2024
40% taking novel therapies (follow up data)

Since 1999, Instituto Nacional de Saúde INSA has been coordinating the Portuguese Familial Hypercholesterolemia (FH) Study, a vital initiative aimed at identifying the genetic cause of hypercholesterolemia—high cholesterol levels present from birth—within families. This work helps reduce cardiovascular risk through early diagnosis and appropriate treatment.

Key Facts About FH in Portugal

Approximately 33,000 people have heterozygous FH, and 25 people have homozygous FH.

Currently, only 3% of people with FH are identified.

Study Achievements (1999–2024)

3,775 cases referred, including:
    1,386 index cases with clinical suspicion of FH
    2,389 relatives

1,150 cases genetically confirmed:
    1,129 heterozygous FH
    15 homozygous FH

40% of identified patients are receiving novel therapies (follow-up data).

This progress underscores the importance of continued collaboration and data sharing to improve FH detection and treatment across Portugal.

Contributed by Prof Mafalda Bourbon

Familial Hypercholesterolemia (FH): Why Awareness and Registries Matter

As part of the 10th Congress of Cardiologists in Bosnia and Herzegovina, held in Sarajevo, a special session titled "Familial Hypercholesterolemia: Why Awareness and Registries Matter" highlighted the urgent need for FH recognition and registry development. This event was organized within the framework of the EAS FHSC project and FH Awareness Week, aiming to raise awareness of FH and its role in premature cardiovascular disease in the region.

Key Highlights from the Session

- The session brought together lipid specialists, cardiologists, laboratory experts, and young clinicians from across the country.
- Free cholesterol and triglyceride screening was offered during the congress, attracting strong interest—over 150 delegates underwent on-site testing.
- This initiative not only raised awareness but encouraged physicians to integrate early FH detection into clinical practice.

Why Registries Matter

A central message was the importance of establishing a national FH registry in Bosnia and Herzegovina. Currently, data are maintained within the EAS FHSC collaboration in an Excel-based format, covering more than 2,800 dyslipidemic patients, with approximately 12% genetically or clinically confirmed FH cases. However, genetic testing is not reimbursed by health insurance, underscoring the need for stronger institutional support.

Call to Action

The session concluded with a call for enhanced collaboration among cardiologists, endocrinologists, and laboratory medicine experts to identify index cases and cascade screen families. Awareness initiatives like this show that even modest resources can make a meaningful impact when combined with professional commitment and education.

This event also marked the 30th anniversary of the Association of Cardiologists of Bosnia and Herzegovina, symbolizing a shared commitment to prevention, knowledge, and solidarity in the fight against FH.

Contributed by Prof. Belma Pojskić, Bosnia and Herzegovina

CEDRA: A National Network for Rare Dyslipidemias and HoFH Care in France

In 2023, the Center of Expertise for Rare Dyslipidemias (CEDRA) was officially certified by the French Ministry of Health and Prevention, marking a major step forward in specialized care for patients with rare lipid disorders, including Homozygous Familial Hypercholesterolemia (HoFH).

CEDRA Network

A multidisciplinary network provides specialized care for both adults and children, ensuring comprehensive management of rare dyslipidemias.

CEDRA consists of: Coordinating Center: Marseille (Pr Sophie Béliard); Eight Expertise Centers: Dijon (Pr Bruno Vergès), Lille (Dr Cécile Yelnik), Lyon (Dr Sybil Charrière), Nantes (Pr Bertrand Cariou), Paris La Pitié Salpétrière (Dr Antonio Gallo), Paris Trousseau (Dr Julie Lemale), La Réunion (Dr Mohammad Ryadh Pokeerbux), Toulouse (Pr Jean Ferrières)

French FH Registry – REFERCHOL

CEDRA actively contributes to REFERCHOL, the national FH registry, which currently includes 185 HoFH patients, including pediatric cases. The registry collects clinical and biological data, supporting research and improving patient care.

Advancing Genetic Diagnosis

CEDRA offers whole genome sequencing using advanced techniques and collaborates with expert genetic clinicians to ensure accurate interpretation, strengthening early and precise diagnosis.

Research and Innovation

CEDRA is deeply involved in academic and clinical research, including the development of innovative therapies. In 2024, members published promising results on evinacumab in American Journal of Preventive Cardiology (Béliard et al., PMID: 38695169):

- 12 HoFH patients (aged 12–57) treated across 3 centers
- 56% LDL-C reduction at 6 months, sustained over 3.5 years
- Zero cardiovascular events in treated patients vs 13 events in controls over 4 years

This breakthrough underscores the transformative potential of novel therapies for HoFH.

Premarital FH Strategy: A New Hope for Families

Homozygous Familial Hypercholesterolemia (HoFH)—the most severe form of FH—poses a serious threat to current and future generations. In many developing countries, limited access to novel lipid-lowering therapies recommended for HoFH patients has left families with few options and little hope.

Recognizing this challenge, we realized it was time to think differently and create a strategy that offers a way forward for the next generation. In 2020, we designed a unique approach titled:
“Premarital Familial Hypercholesterolemia: A New Hope for Families.”

This groundbreaking work, co-authored by Prof. Raul Santos, was recently published in the September issue of the American Journal of Preventive Cardiology. The strategy provides premarital couples three options to prevent having a child affected by this life-threatening genetic lipid disorder. We believe that with correct implementation, this approach can truly bring light at the end of the tunnel for families worldwide. Link of the Article

Updates from the Japan Atherosclerosis Society (JAS): FH Academy and October Highlights

The Japan Atherosclerosis Society **(JAS)** successfully hosted the third FH Academy in Tokyo on September 27, welcoming 50 participants. The program featured a comprehensive curriculum, including:

- Genetic testing and pediatric FH screening
- Advanced imaging for severe FH
- Sex differences and perinatal management
- Latest updates on lipid-lowering therapies

Participants engaged in hands-on clinical training, such as observing corneal arcus, palpating tendon xanthomas, taking detailed family histories, and performing Achilles tendon ultrasounds on HoFH patients. Two FH patients shared their personal journeys, and a previous Academy participant described how the program inspired the creation of a new clinical system at his hospital

October Events: Raising Awareness Nationwide (October 18 – FH Awareness Day Seminar in Hirosaki)

Organized in collaboration with the Research Group on Intractable Diseases in Dyslipidemia (Ministry of Health, Labour and Welfare), attracting nearly 100 participants. Highlights included:

- New insights from the Tohoku Medical Megabank on FH gene variants in the general population.
- Emphasis on involving local governments in pediatric FH screening initiatives.

October 27 – FH Patient Gathering in Osaka

This event brought together 60 participants, including patients and families, fostering community support, Attendees included:

- 10 key opinion leaders
- 13 representatives from pharmaceutical and medical device companies
- 2 officials from the Osaka Prefectural Government
- Opening remarks were delivered via Zoom by Dr. Hanako Jimi, physician and member of the House of Councilors, setting an inspiring tone.

Driving FH Innovation in Western Australia: EDIFICE, FH in KIDS & FH Australia

Western Australia continues to lead the way in improving the identification and management of Familial Hypercholesterolemia (FH) through three major initiatives:

EDIFICE – Enhancing Detection of FH in General Practice

EDIFICE tackles the low uptake of genetic cascade testing, a cost-effective strategy recommended by expert guidelines. The initiative emphasizes the critical role of general practice (GP) in FH detection, supported by:

- Insights from national implementation science workshops
- Australian Government funding for genetic testing

  The core hypothesis: a tertiary-initiated, shared-care model can effectively identify new FH cases in at-risk families. EDIFICE focuses on first-degree relatives and younger patients, with service and clinical outcomes reported in a revised manuscript currently under review.

FH in KIDS – Redefining FH as a Paediatric Disorder Led by Perth Children's Hospital, FH in KIDS targets early diagnosis in children. Despite FH affecting 1 in 250 people (around 2,000 children in WA), fewer than 3% were diagnosed when the program began. Ambitious goal: Identify up to 25% of all children with FH in WA within three years through:

- Cascade testing of relatives in collaboration with Royal Perth Hospital
- Enhanced referral processes and GP engagement
- Expanded clinic services, including Australia's first paediatric FH nurse practitioner
- Early treatment ensures children can achieve normal life expectancy.

FH Australia – Advocacy and Awareness

FH Australia, the national patient-led voice, delivered a week-long awareness campaign around Global FH Awareness Day (September 24).

Highlights:

- Support from seven hospitals across Perth and Sydney
- Point-of-care cholesterol testing and community education
- Free heart health snapshot reviews, attracting strong public interest
- FH Australia continues to advocate for better detection and diagnosis nationwide.
- Learn more: FH Australia Website

Contributors: Prof Gerald F Watts, Christine Tawtel (Hendricks), Dr Jing Pang & Dr Andrew Martin

World Heart Day in Zimbabwe: A Historic Milestone for FH Care

The Launch of Mpilo Lipid Clinic

Mpilo Hospital and Brave Little Hearts in collaboration with IAS and ILCN celebrate the official launch of the first Lipid Clinic in Zimbabwe.

On September 29, 2025, the International Atherosclerosis Society (IAS) celebrated World Heart Day in Zimbabwe in a truly distinguished way!
After months of collaboration with colleagues at Mpilo Hospital in Bulawayo, and with outstanding coordination by the Brave Little Heart Foundation, we proudly launched the first-ever Lipid Clinic in Zimbabwe.

Building the Foundation

Prior to this landmark event, a series of meetings and in-depth discussions were held on:
The importance of early detection and proper treatment of Familial Hypercholesterolemia (FH)—a genetic lipid disorder that significantly shortens life expectancy.
The need to implement screening programs for affected families.
These efforts culminated in a dedicated FH Symposium for hospital staff, summarizing best practices and global strategies for FH management.

Why This Matters

This achievement marks another success for the global FH community, demonstrating our commitment to supporting colleagues in regions of unmet need. By establishing specialized clinics and promoting awareness, we can ensure early diagnosis and effective treatment for patients worldwide.

Contributed by Dr Mutaz Al-Khnifsawi Iraq

NEWS FROM FH EUROPE FOUNDATION

SHORT COMMUNICATION · Articles in Press, 120447, August 19, 2025

Open Access

atherosclerosis

Lp(a) testing for the primary prevention of cardiovascular disease in high-income countries: a cost-effectiveness analysis

A new international study reveals that routine testing for lipoprotein(a), or Lp(a), could prevent heart attacks, strokes, and save millions in healthcare costs. Lp(a) is a genetic, lifelong cardiovascular risk factor affecting nearly 1 in 5 adults, yet it remains largely overlooked in routine screenings.

Led by Professor Zanfina Ademi at Monash University, the study highlights that even individuals with healthy lifestyles and normal cholesterol levels may still face serious risk if their Lp(a) is elevated. Unlike cholesterol, Lp(a) levels cannot be lowered through diet or exercise.

Published in *Atherosclerosis*, the study analysed data from over 10,000 UK adults. Routine testing would have reclassified 20% as high-risk, enabling earlier intervention with blood pressure or cholesterol-lowering treatments. The modelling showed that per 10,000 people tested, Lp(a) screening could prevent 60 heart attacks, 13 strokes, and 26 early deaths—while adding hundreds of healthy life years and saving approximately $85 (Australia) and £263 (UK) per person in medical and productivity costs.

Experts including Prof. Florian Kronenberg and Magdalena Daccord (FH Europe) call for urgent policy action. The findings informed the Brussels International Declaration on Lp(a), a global roadmap advocating for its inclusion in national cardiovascular screening programs.

Prof. Jan Boren, Editor-in-Chief of *Atherosclerosis*, emphasized: “When a simple blood test can prevent life-threatening events and improve quality of life, implementing it is imperative.”

Commissioned by the Lp(a) International Taskforce and hosted by FH Europe Foundation, the study was a patient-driven initiative led by Monash University’s Centre for Medicine Use and Safety, in collaboration with global researchers. It marks a major step forward in addressing one of heart health’s biggest blind spots.

UPCOMING EVENTS

EAS
Paediatric Lipid Working Group

SAVE THE DATE

The 9th EAS Paediatric Familial Hypercholesterolemia Symposium

Saturday, 23 May 2026 | Athens, Greece

Topics:

Dietary-nutritional treatment of FH: Mediterranean diet, Nordic diet, nutraceuticals

Additional therapies for children with HeFH when LDL-C is not at target or when Lp(a) is high

Childhood Universal Screening strategies for FH – What steps are needed to ensure maximum possible benefit and minimal potential harm?

EAS
94th Congress
24-27 May 2026 | Athens, Greece

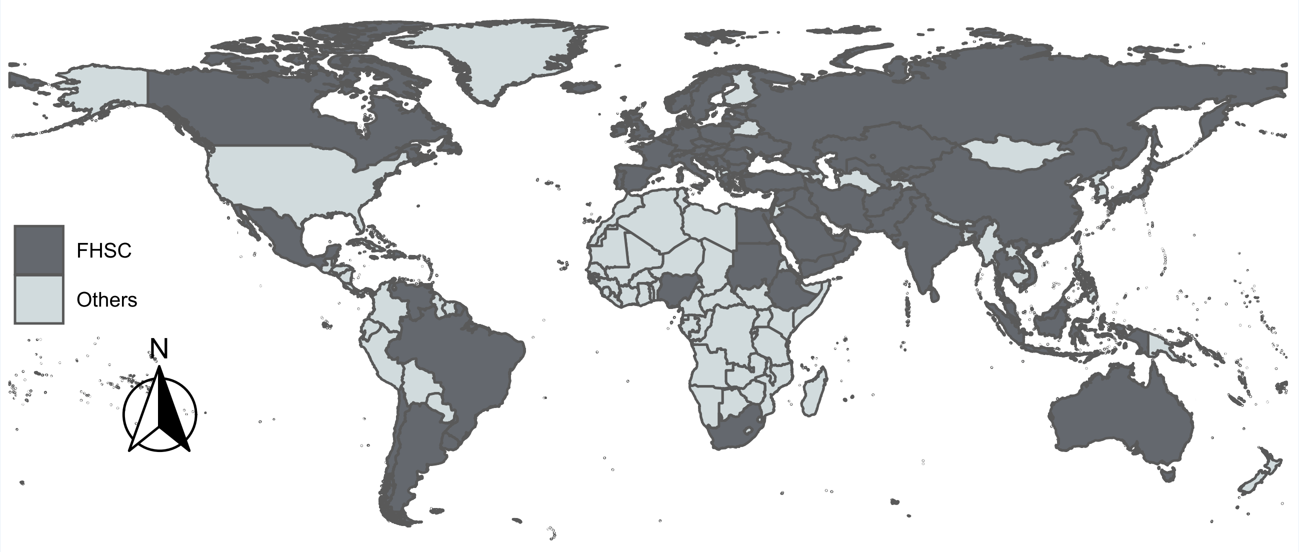
Organised by: Christina Katsagoni, Uma Ramaswami, Jeanine Roeters van Lennep, Noel Peretti, Urh Groselj, Mafalda Bourbon, Kirsten Holven, Michal Vrablik, Evangelos Liberopoulos & Steve Humphries

The EAS Congress next year and will take place from 24-27 May 2026.

The EAS FHSC
Steering Committee Closed Meeting
will be held on the morning of
Sunday 24 May 2026.

- By invitation only -
- Limited to approx. 100 attendees -

GET INVOLVED



FHSC spans 76 countries and includes 92 National Lead Investigators.
The FHSC Registry includes approx. Approx. 85K cases across 69 countries.

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 (NCT04272697)? If so, we would like to hear from you! For enquires contact info@eas-fhsc.org.

More information 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

The 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.

Worldwide Directory of Lipid Clinics & Patient Support Groups:
findmylipidclinic.com For more information: Stevens *et al. EAJ* 2022;2:37-40