FHSC
EAS Familial Hypercholesterolaemia Studies Collaboration

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NEWSLETTER

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FHSC
EAS Familial Hypercholesterolaemia Studies Collaboration

FEATURED ARTICLE

FHSC Publication in the European Heart Journal

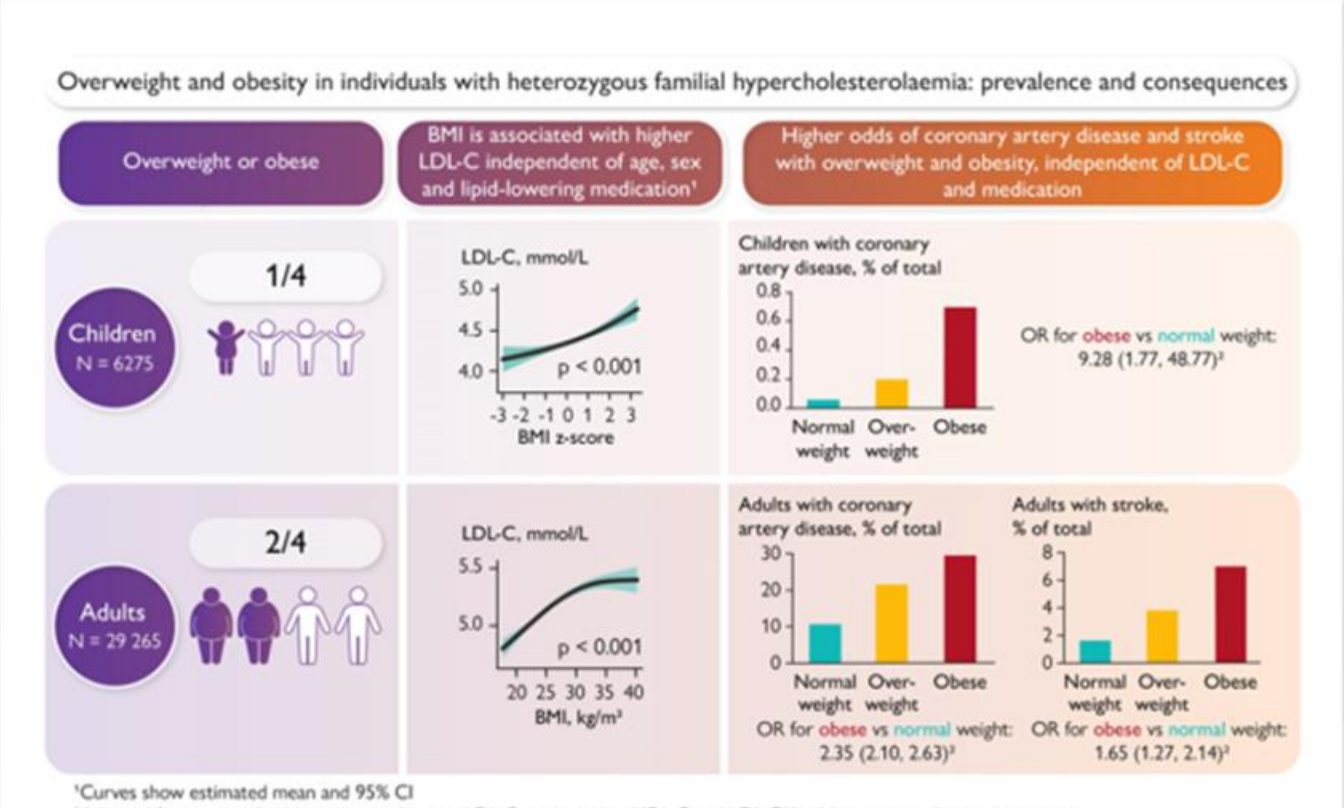
Overweight, obesity, and cardiovascular disease in heterozygous familial hypercholesterolaemia: the EAS FH Studies Collaboration registry

European Heart Journal | ESC European Society of Cardiology

➔ Article Link

The Coordinating Centre is delighted to announce our recent publication exploring the global prevalence of obesity in patients with FH and its impact on cardiovascular disease risk.

This work celebrates the dedicated collaboration of FHSC investigators from 50 countries who shared baseline data between 2015 and 2022.



Overweight and obesity in individuals with heterozygous familial hypercholesterolaemia: prevalence and consequences

Overweight or obese

BMI is associated with higher LDL-C independent of age, sex and lipid-lowering medication[1]

Higher odds of coronary artery disease and stroke with overweight and obesity, independent of LDL-C and medication

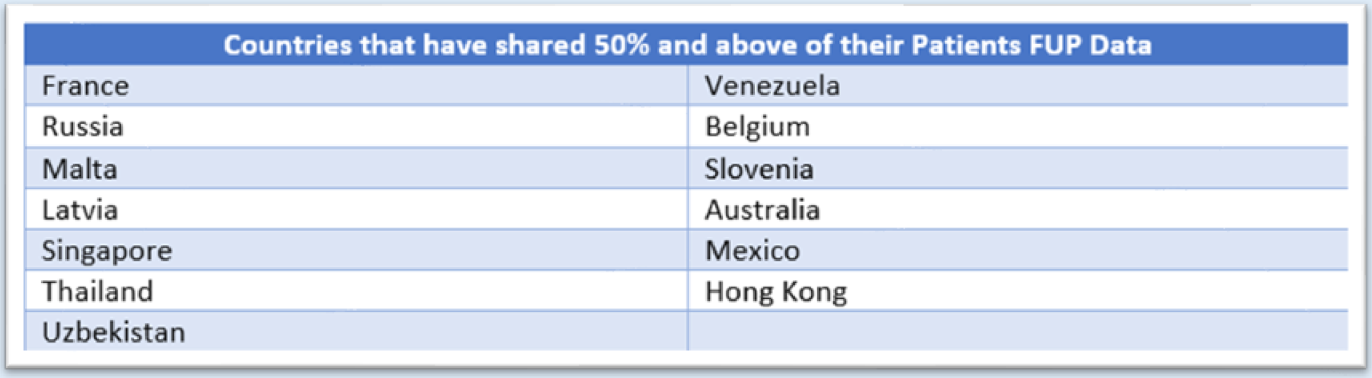
[1] Curves show estimated mean and 95% CI

[2] Adjusted for age, sex, lipid-lowering medication, LDL-C, triglycerides, HDL-C and LDL-C*lipid-lowering medication interaction

COORDINATING CENTRE'S UPDATE

Call for Follow up Data: A Global Effort

We extend our call for follow-up data to the countries that have not yet shared their information with the FHSC Coordinating Centre. As of this newsletter's publication, 25 countries have responded to the call made in November 2024 by sharing their data with us. The table below summarizes the number of countries that have managed to share follow-up data and outcomes for more than 50% of their patients.



Countries that have shared 50% and above of their Patients FUP Data	
France	Venezuela
Russia	Belgium
Malta	Slovenia
Latvia	Australia
Singapore	Mexico
Thailand	Hong Kong
Uzbekistan	

Sharing these datasets is crucial for the Coordinating Centre to understand how FH is managed globally and to identify the risks associated with this genetic condition.

We greatly appreciate the efforts of all National Lead Investigators in sharing these datasets and responding to our calls.

The final date to share these datasets is **May 31, 2025**. We invite NLIs who have not yet shared follow-up data to upload their datasets or enter their patients' information on the FHSC Web Portal.

Kazakhstan Launches First National Registry for FH

Professor Davletov Kairat, National Lead Investigator for Kazakhstan, has launched the country's first FH registry. As Principal Investigator, he leads the project "Development and implementation of the national registry of patients with FH in the Republic of Kazakhstan" (ИРН AP19680286), funded by the Ministry of Education of the Republic of Kazakhstan. The project aims to establish a comprehensive national registry to enhance monitoring and treatment of FH patients, while investigating demographic and genetic factors associated with the condition. The intermediate results of the project were recently presented at the first Congress of the Kazakhstan Atherosclerosis Society, which is dedicated to advancing lipidology and improving patient care to mitigate cardiovascular diseases in Kazakhstan. This milestone event provided an excellent platform to discuss the progress of this initiative and foster collaborations.

Novel Mutation in Indian Family Highlights Importance of Early FH Detection

Dr Tester F. Ashavaid, National Lead Investigator for India, and her team from P. D. Hinduja Hospital and Medical Research Centre, have recently published an article in the Indian Journal of Clinical Biochemistry.

Their study, entitled 'Likelihood of a Novel Pathogenic LDLR Mutant Allele Inherited Preferentially within an Indian Family with Familial Hypercholesterolemia', identifies a previously unreported nonsense mutation in the LDLR gene (c.743_744delinsAA, p.Cys248*) in a Mumbai-based family. This discovery is particularly significant as it reveals an unusual inheritance pattern of the mutant allele and highlights the importance of cascade screening in identifying at-risk family members, especially in populations where genetic data on FH remains limited.

Indian Journal of Clinical Biochemistry

Association of Clinical Biochemists of India

Springer

Article Link

Successful 9th FH Week and New National Cardiovascular Plan in Czechia

The Czech Republic's annual "FH Week", established in 2015, coincides with World Familial Hypercholesterolemia Day each September. Organised by the Czech Alliance for Cardiovascular Diseases (CAKO) with support from the Czech Society for Atherosclerosis and Medped project, the initiative provides on-site health screenings including lipid panels and refers potential FH cases to specialist lipid centres. In 2024, the programme screened over 700 people, identifying eight new FH families and conducting a pilot Lp(a) testing programme where 20% of participants showed at-risk values. Since inception, approximately 50 FH families have been identified. Additionally, in late 2024, the Czech Republic became Europe's third country to implement a National Cardiovascular Plan, which notably includes universal paediatric FH screening and Lp(a) measurement in primary care. Read More

30.000 Children Screened for FH in Germany: the VRONI Study

The VRONI study, launched with the slogan "Heart attack at age 35? Without me!", aims to detect and treat familial hypercholesterolemia (FH) early in children and adolescents. By December 2024, the study had screened 30,000 children in Bavaria. Children aged 5 to 14 are tested during routine paediatric visits, with genetic testing conducted if LDL cholesterol exceeds 130 mg/dl (3.42 mmol/l). The study has identified 265 FH cases, with positive cases receiving specialist care. Following its success, VRONI has expanded to Northern Germany, now involving over 800 doctors. The programme implements reverse cascade screening to identify affected family members and provides online genetic counselling. Despite its achievements in improving FH detection and care, political challenges continue to hinder nationwide implementation.

Hellas FH Registry: Advancing FH Research in Greece

The Hellenic Familial Hypercholesterolemia (Hellas-FH) Registry has completed 8 years of progress, enrolling over 3700 patients across 25 sites in Greece. At the 2024 National Congress of the Hellenic Atherosclerosis Society, two projects were presented: an initiative to identify lysosomal acid lipase deficiency (LAL-D) cases often misdiagnosed as FH, using specific diagnostic criteria; and the development of HELp(a), a registry for elevated lipoprotein(a) levels, to better understand Lp(a)-related cardiovascular disease. These achievements reflect the collective efforts of all investigators, to whom we extend our gratitude. In 2024, Hellas-FH publications covered key FH topics: obesity and CVD, physical signs and ASCVD, lipid-lowering treatment and MASLD, and triglycerides and ASCVD (in press).

National Lead Investigators interested to contribute an article in our quarterly Newsletter?
(e.g. FH news, publications, events)
Please contact: info@eas-fhsc.org

FH-EARLY: A Bold Leap Toward Revolutionising Diagnosis and Co-Management of Familial Hypercholesterolemia

The FH-EARLY project, launched on January 1, 2025, is a major step in precision medicine and patient-centered care for familial hypercholesterolemia (FH). Funded with €7.25 million under the HORIZON Research and Innovation Actions, it runs until December 31, 2028. The initiative, led by AIDFM (Portugal) and involving 15 institutions from Europe, Lebanon, and Turkey, tackles the underdiagnosis and poor management of FH, which affects over 30 million people worldwide, yet fewer than 10% are diagnosed.

The project integrates genomics, AI, and bioinformatics to improve early diagnosis and treatment, particularly for young individuals (16-24 years). Key innovations include:

- Next-Generation Chip Array for cost-effective genetic testing
- Signature Biomarker for better risk assessment
- AI-Driven Precision Health Profiling (iPHP) Tool for FH management

Beyond medical advancements, FH-EARLY promotes health literacy, emotional resilience, and better communication, focusing on underserved populations. By enhancing early detection and personalized care, the project aims to reduce cardiovascular disease (CVD) costs in the EU and support the development of a Cardiovascular Health Plan, ultimately improving long-term health and saving lives. Read More

FHSC NLIs Among the World's Most Cited

The FHSC Coordinating Centre extend their heartfelt congratulations to the National Lead Investigators who have been named in the 2024 Clarivate Highly Cited Researchers list. Each year, Clarivate recognizes academic authors whose publications have garnered a significant number of citations in academic journals. These distinguished researchers represent approximately 0.1% of the global scientific community.

Having our National Leads members included in this elite group underscores our commitment to publishing high-impact research. It also reinforces our mission to empower the medical and global community to advocate for changes in the detection, management, and clinical outcomes of FH. Our goal is to promote early diagnosis and more effective treatment of FH worldwide.

UPCOMING EVENTS

EAS

93rd Congress

4-7 May 2025 | Glasgow, UK

Save the Date!

EAS European Atherosclerosis Society

eas-congress.com/2025

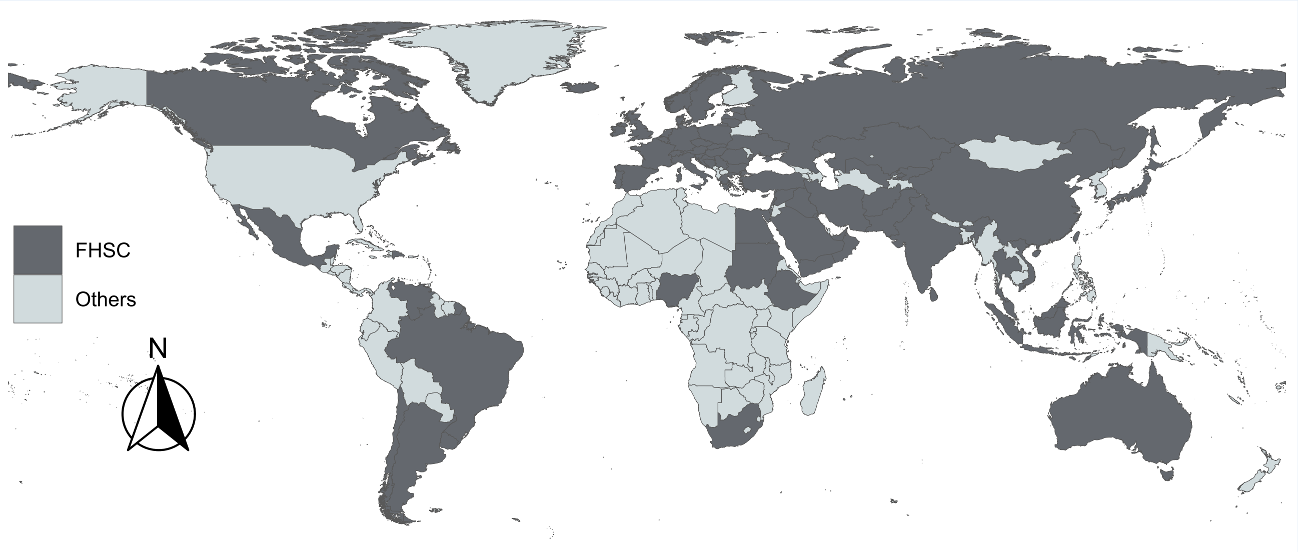
The EAS Congress is one month earlier this year and will take place from 4-7 May 2025.

The EAS FHSC
Steering Committee Closed Meeting
will be held on the morning of
Sunday 4 May 2025.

- 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. 80K cases across 68 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