FHSC

EAS Familial Hypercholesterolaemia Studies Collaboration

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NEWSLETTER

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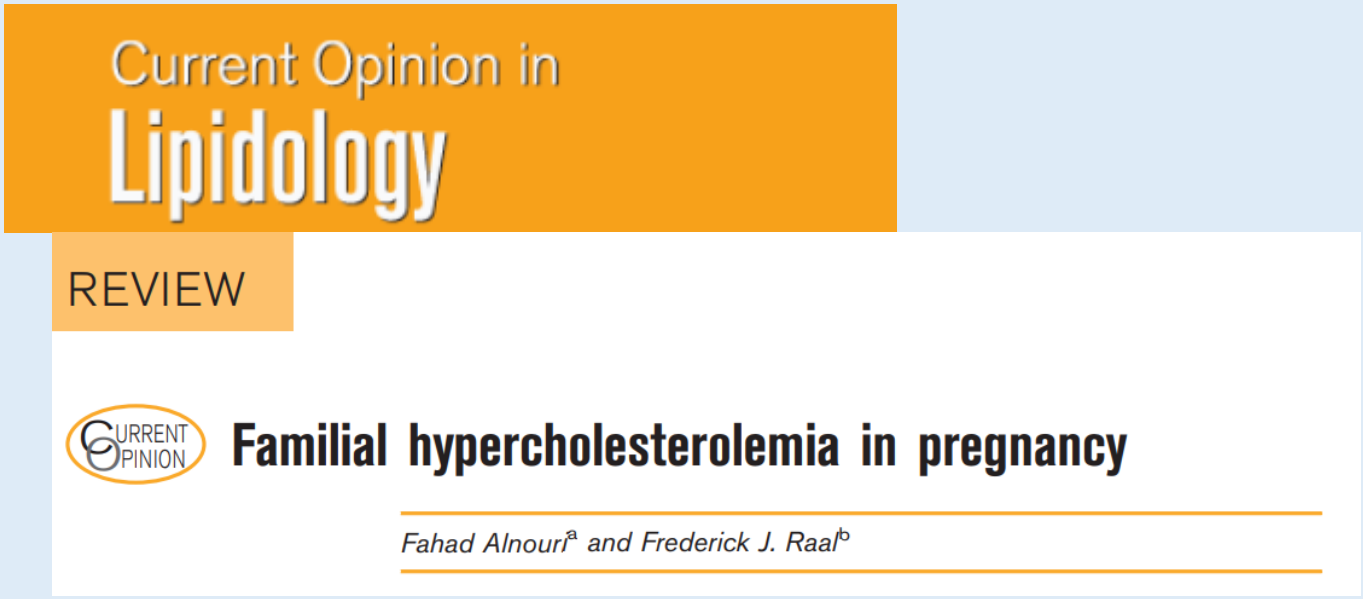
Hypercholesterolemia in Pregnancy

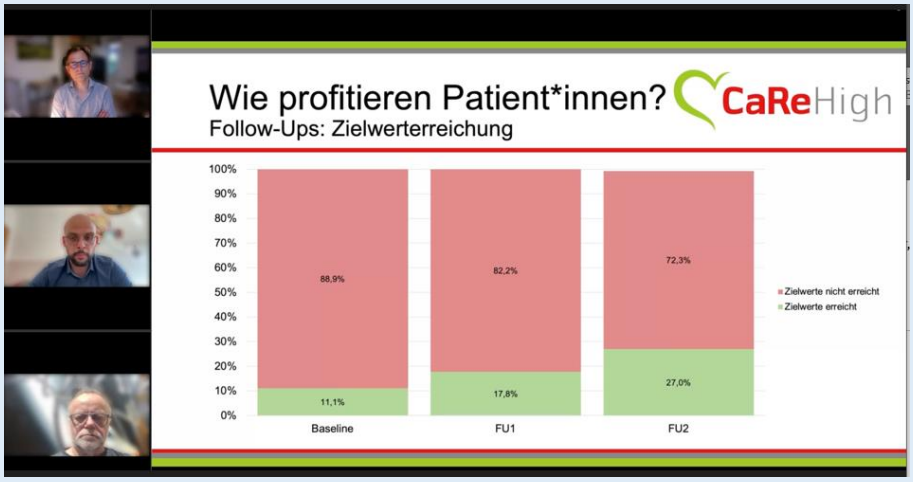


Current Opinion in Lipidology: Link

Profs Fahad Alnouri and Derick Raal (NLIs for Saudi Arabia and South Africa) have authored a review examining familial hypercholesterolemia (FH) management during pregnancy, particularly homozygous FH (HoFH). Patients with HoFH face markedly elevated LDL-cholesterol from birth and present with unique complications at pregnancy. Despite FH's greater burden than previously thought, underdiagnosis persists and evidence-based pregnancy guidelines for HoFH remain absent.

Management requires balancing maternal LDL-cholesterol control with foetal safety, potentially discontinuing lipid-lowering medications during pregnancy and the postpartum period needs to be considered, and in severe cases, lipoprotein apheresis may be an appropriate substitute. Multidisciplinary care involving genetic counsellors, cardiologists, lipidologists, and obstetricians is essential. Enhanced research, public awareness, and international collaboration are needed to improve outcomes for pregnant women with HoFH.

German CaRe High Registry Kicks Off Patient Webinar Series with Strong Engagement



In July 2025, **CaRe High** — the German registry for patients with familial hypercholesterolemia (FH) — hosted its very first patient webinar with outstanding success. More than **220 participants** joined this premiere event, a response that far exceeded expectations and highlighted the strong interest of patients in learning more about their condition.

The program featured insightful presentations by key members of the CaRe High team. **Prof. Winfried März,** principal investigator of the study, provided a comprehensive overview of the genetic background of FH. **Felix Fath,** team and project lead, presented key findings from the CaRe High registry and introduced the newly developed **CaRe High patient app,** designed to support patients in managing their condition. Finally, **Philipp von Gallwitz** from Partners4Patients spoke about the chance of founding a patient organization, aiming to strengthen the patient community and advocacy efforts.

However, the highlight of the event came unexpectedly: an overwhelming number of questions — more than **70 inquiries** — were submitted by participants, covering topics such as medication, side effects, app usage, and the interpretation of individual lipid profiles. Due to this remarkable engagement, the CaRe High team spontaneously organized a **second follow-up session,** dedicated to answering patient questions. **This Q&A session was offered exclusively to registered participants of the first webinar,** and still attracted **over 100 attendees,** once again demonstrating the strong demand for dialogue and personalized information.

These experiences have clearly shown the energy and commitment that comes directly from the patient community. Motivated by this response, **CaRe High will now offer 3 to 4 webinars annually,** creating regular opportunities for patient education, exchange, and empowerment.

We thank all participants for their trust and look forward to building a vibrant and active community together.

Contributed by **Prof. Winfried März – National Lead investigator Germany**

HELLAS-FH Registry: Advancing Familial Hypercholesterolemia Care in Greece

Founded in 2016 under the Hellenic Atherosclerosis Society, the HELLAS-FH Registry is a nationwide initiative focused on improving diagnosis, care, and understanding of familial hypercholesterolemia (FH) in Greece. It now includes 26 centers across seven major cities, with nearly 4,000 patients enrolled—about 10% of the country's estimated FH population.

HELLAS-FH is expanding strategically, with new sites joining and patient recruitment increasing. A major milestone is the longitudinal tracking of over 1,260 patients, providing valuable real-world data on FH management. The registry continues to grow, promoting broader access to diagnosis and treatment.

Academically, HELLAS-FH is active in national and international congresses, raising awareness among professionals and patients. It collaborates with FH patient groups to ensure outreach is patient-centered and impactful.

The registry's scientific contributions include over a dozen peer-reviewed publications on LDL-C targets, cardiovascular risk, and metabolic conditions. Recent studies have explored links between FH and liver function, triglycerides, and hypertension.

- *Boutari C. et al.* – The effect of lipid-lowering treatment on indices of MASLD in FH patients. *J Cardiovasc Med*, 2024.
- *Anagnostis P. et al.* – Correlation between triglyceride levels and ASCVD in adults with FH. *J Clin Lipidol*, 2025.
- *Antza C & Rizos CV. et al.* – Unraveling the hypertensive factor in FH. *J Hum Hypertens*, 2025 (in press).

Beyond research, HELLAS-FH is committed to education and advocacy. Its website (www.hellasfh.gr) and social media (@hellasfh on X, Hellas FH on Facebook) serve as platforms for public engagement and professional learning. The registry also works with health authorities to shape national cardiovascular prevention policies.

During FH Awareness Week, the registry emphasized the life-changing importance of early detection and treatment. Through ongoing efforts, HELLAS-FH is advancing equitable care, reducing cardiovascular risk, and improving lives across Greece.

Hungarian FH Registry Advances Awareness and Treatment Strategies

Since its establishment in **2016**, the **Hungarian Familial Hypercholesterolemia (FH) Registry** has been working to improve diagnosis and care for individuals with FH. Currently, **18 centers** across the country contribute to the registry, which uses the **Dutch Lipid Clinic Network criteria** for diagnosis.

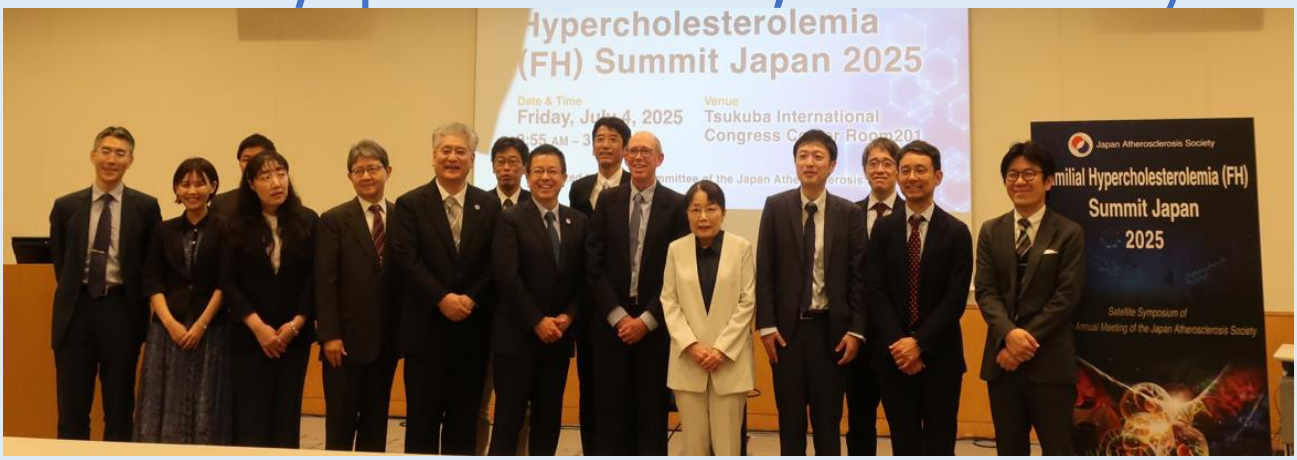
To date, **459 patients** have been enrolled, including **six with the homozygous FH phenotype**.

To expand understanding and raise awareness, the registry team applied a **data mining approach** to analyze records from **1.3 million individuals with cardiovascular disease**, estimating FH likelihood based on Dutch criteria. Treatment strategies include **statins, ezetimibe, and PCSK9 inhibitors**, aligned with **2019 EAS/ESC LDL-C targets**. Patients begin with a **maximally tolerated statin**, followed by **statin + ezetimibe** if targets are unmet, and **PCSK9 inhibitors** as a third-line option. Notably, only a small percentage of patients currently receive the statin-ezetimibe combination. For those not reaching LDL-C targets, **LDL apheresis** is also utilized.

The registry actively participates in **FH Awareness Week**, organized by the **EAS FHSC**, and regularly engages the public through **national and local media** to promote FH awareness. FH-related findings have been presented at **domestic and international congresses**, and several **publications** have been produced (attached).

In addition, **Lp(a) testing** is routinely performed at lipid specialist clinics and cardiology centers. Data from general practitioners and institutional assessments are partially published and ongoing. Current therapies aimed at reducing the high absolute risk in individuals with elevated Lp(a) focus on more aggressively lowering LDL-C levels, beyond statins, including the use of PCSK9 inhibitors.. Currently, **high Lp(a)** is **not an indication for LDL apheresis** in Hungary. The registry remains committed to improving FH care, raising awareness, and contributing to scientific research and public health advocacy.

Japan Atherosclerosis Society Advances FH Care Through Summit, Updated Guidelines, and FH Academy



On **July 4, 2025**, the **Japan Atherosclerosis Society (JAS)** hosted the **FH Summit Japan 2025** in Tsukuba as a Satellite Symposium of its **57th Annual Scientific Meeting**. The event brought together leading international experts who shared cutting-edge research and clinical insights on key topics such as intractable FH, homozygous FH (HoFH), FH diagnosis and treatment in East Asia, genetic approaches, and innovative studies led by emerging young researchers. The summit was a resounding success, marked by dynamic presentations and vibrant discussions that fostered international collaboration.

FH-related sessions were also featured throughout the 57th Annual Meeting, further underscoring JAS's commitment to advancing FH research and clinical care.

In **June 2025**, JAS released the **Guidelines for the Diagnosis and Treatment of Familial Hypercholesterolemia – Focused Update 2025** on its official website, coinciding with the launch of new therapies for HoFH. The update provides the latest recommendations for treating HoFH in both adults and children. *(Note: Available in Japanese only.)*

Looking ahead to **September**, JAS will host the **third FH Academy**, a hands-on training program focused on FH diagnosis and treatment. The program includes a unique opportunity to hear directly from a patient living with FH. In addition to lectures, participants will gain practical experience in clinically relevant skills such as observing corneal arcus, palpating tendon xanthomas, conducting detailed family history assessments, and performing Achilles tendon ultrasound evaluations. This annual seminar continues to receive high praise for its comprehensive and practice-oriented approach.

Furthermore, this **autumn**, in conjunction with **FH Awareness Day**, JAS will organize a special seminar and patient gathering. These events are held in collaboration with the **Research Group on Intractable Diseases in Dyslipidemia** (sponsored by the Ministry of Health and Welfare) and the **Patients' Association for Refractory Familial Hypercholesterolemia**. The initiative aims to foster patient engagement, strengthen community connections, and serve as a vital platform for raising public awareness and promoting a deeper understanding of FH among healthcare professionals and the general public.

Multiple Initiatives to Assess the Prevalence of Familial Hypercholesterolemia in Pakistan

Familial hypercholesterolemia (FH) remains significantly underdiagnosed in Pakistan. Our team has undertaken several large-scale initiatives to better understand the prevalence of FH, gender disparities in lipid testing, and dyslipidaemia patterns across various age groups—including among children.

In a recent study, we analyzed lipid profile data from over 500,000 individuals across two major laboratory networks in Pakistan to estimate the national prevalence of FH (1). By integrating these findings with data from an earlier Pakistani study, we derived a pooled FH prevalence estimate of 1 in 273 among 1.5 million individuals. Strikingly, the highest prevalence was observed in individuals under 20 years of age (1 in 29), highlighting the urgent need for early detection and targeted screening strategies. These findings were presented at the 20th International Symposium on Atherosclerosis (ISA 2024), hosted by the International Atherosclerosis Society (IAS).

We also examined gender disparities in adult lipid testing, discovering that men were more frequently tested at younger ages, while women were typically tested later in life despite exhibiting higher levels of LDL-C, total cholesterol (TC), and HDL-C (2). These results emphasize the need to promote earlier screening among women to ensure timely diagnosis and management of lipid disorders.

Our recent paediatric research revealed a high burden of dyslipidaemia among children and adolescents in Pakistan, with many showing elevated cholesterol, low HDL-C, and high triglyceride levels (3). The findings also exposed gender and geographic disparities—boys were disproportionately affected, and testing rates varied by region. These observations reinforce the necessity of equitable, nationwide screening programs for children.

Collectively, these studies highlight the urgent need for widespread screening initiatives and public awareness campaigns targeting FH and dyslipidaemia across all age groups in Pakistan.

1. Khan M, Ain Q, Sikonja J, Batool H, Hayat MQ, Khan MI, et al. Prevalence of Familial Hypercholesterolemia in Pakistan: A Pooled Analysis of 1.5 Million Individuals and Comparison with Other Countries of the Region. Glob Heart [Internet]. 2025 Mar 10;20(1):23. Available from: https://account.globalheartjournal.com/index.php/up-j-gh/article/view/1413
2. Nawaz A, Khan M, Ain Q, Amjad M, Sikonja J, Batool H, et al. Gender Disparity in Lipid Testing Among Over 0.5 Million Adults from Pakistan: Females are Tested Much Later Despite Higher LDL-Cholesterol Levels. Glob Heart [Internet]. 2025 Feb 21;20(1):16. Available from: https://account.globalheartjournal.com/index.php/up-j-gh/article/view/1401

Participation in Proprotein Convertase Subtilisin/Kexin Type 9 (PCSK9) Inhibitor Research Study

Dr Mary Abba and Prof Krizstof Chlebus, from Gdansk invite healthcare practitioners to actively participate in a clinical research study examining the biochemical effects of PCSK9 inhibitor therapy in patients with hypercholesterolemia or established cardiovascular disease.

The study aims to assess the biochemical profiles of patients currently receiving PCSK9 inhibitors (e.g., alirocumab, evolocumab) through scheduled blood testing. We are seeking clinicians who are willing to collaborate by recruiting eligible patients from their practices.

Participating practitioners will play a key role in identifying suitable patients, obtaining informed consent, and coordinating biochemical blood testing in accordance with the study protocol.

By participating, you will contribute to a deeper understanding of the real-world effects of PCSK9 inhibitors and help inform future clinical guidelines. This is a valuable opportunity to be part of a collaborative effort that bridges clinical practice and academic research. We welcome your expertise and partnership in this important study.

Please reach out to maia.abba@gumed.edu.pl if you are interested to contribute.

SHORT COMMUNICATION · Articles in Press, 120447, August 19, 2025 atherosclerosis
Open Access

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.

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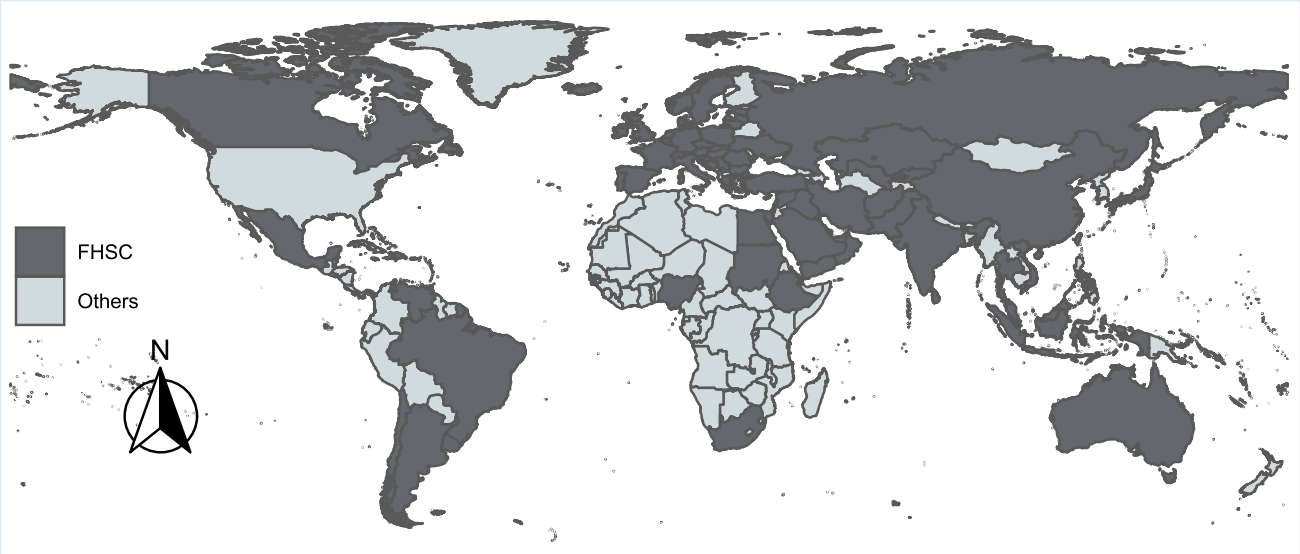
GET INVOLVED



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