Understanding disparities in Familial Hypercholesterolaemia

When we think about what shapes our health, it is easy to focus only on biology. In reality, social and economic factors play a huge role as well. Things like income, education, where we live, and our job quality all influence our health.

This is just as true for *Familial Hypercholesterolaemia (FH)* – an inherited condition that causes high cholesterol levels and increases the risk of early heart disease. Factors beyond our control impact how FH is understood, diagnosed, and treated, causing inequities that we call FH disparities. To prevent FH disparities, we first need to understand them.

Health Gaps in Europe and beyond

Before we can understand FH disparities however, it is important to understand what health inequities look like. Across Europe, there are sharp differences in health that we can identify. For example:

- People on low incomes are far less likely to report being in good health than those on higher incomes;
- Women report poorer health than men;
- Life expectancy can vary by more than 12 years between regions, and the gap is even bigger for minority communities such as Roma.

These inequities are caused by the **social determinants of health**, or the non-medical factors linked to the place or conditions where we are born, grow up, live, work, and age. The Dahlgren-Whitehead rainbow helps us understand the impact of these factors.

Health is shaped by the interaction of broad structural conditions, our living and working environments, social and community networks, and individual lifestyles and behaviours. Age, sex, and genetics also play a role but are factors that cannot be changed.

In this way, these factors also impact FH prevalence and care, causing FH disparities in very specific ways.

Co-funded by the European Union

This project received funding from European Union's EU4Health Programme (EU4H) REF: 101129403 — PERFECTO — EU4H-2022-PJ-3. Views and opinions expressed are however those of the author(s) only and do not necessarily reflect those of the European Union or HaDEA. Neither the European Union nor the granting authority can be held responsible for them.

Who gets left behind in FH care?

The evidence base for FH disparities is limited, but studies from across the world show clear patterns:

- **Access to treatment**: Men who are white, wealthier, and more educated are more likely to be prescribed cholesterol-lowering drugs.
- **Representation in research**: Clinical trials often exclude women, ethnic minorities, and lower-income families, meaning findings do not reflect the full population.
- **Barriers in genetic testing**: Minority groups face barriers to testing due to cost, awareness, mistrust, and access issues.
- **Health literacy inequities**: People with lower income or education levels are less likely to fully understand aspects of FH, or how to manage the condition or its treatment.
- **Gender gaps**: Women are typically diagnosed later than men, start treatment later, and face unique risks during pregnancy and menopause.

When comparing countries, the disparities are even bigger. In lower-income countries diagnosis is less common, fewer people get access to treatment, and cardiovascular risks are higher.

Why does this matter?

These disparities show that certain groups are consistently left out of testing, treatment, and research, worsening their health outcomes. The cycle continues: under-diagnosis leads to under-treatment, fuelling risks of heart disease and early death. This show us that, even if we had a perfect system in place for diagnosing, understanding, and treating FH, there would always be people left out.

What needs to change

Addressing FH disparities requires continuously growing knowledge of what drives them and their impacts. This means an expansion of the current research base, but also the development of concrete solutions to ensure that nobody with FH goes undiagnosed, or without treatment.

The EU4Health project **PERFECTO – Preventing the Preventable: Transforming Cardiovascular Health** aims to prevent FH disparities. Its fourth pillar focuses on equitable paediatric screening for marginalised groups in Romania (Roma communities) and Cyprus (migrant populations). Led by the European Public Health Alliance (EPHA), it collects research on FH disparities to find concrete solutions to apply these to policy and practice in the European Union.

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Body of resources

Curious about research on FH disparities? The resources below provide an overview of the most relevant articles collected for the work of PERFECTO:

- 2009 – Underrepresentation of non-white children in trials of statins in children with heterozygous familial hypercholesterolaemia (Belay, B., Racine, A.D., & Belamarich, P. F)
  - Scope: Global (AT, CA, FI, GR, NL, NO, ZA, US)
  - Findings: Non-white children are severely underrepresented in paediatric statin trials for HeFH; 92% of study participants were white

- 2013 – The Need to Build Trust: A Perspective on Disparities in Genetic Testing (Saulsberry, K., & Terry, S.F)
  - Scope: United States
  - Findings: Minority groups have lower uptake of genetic testing due to location, socioeconomic barriers, lack of awareness, and distrust; highlights need for direct engagement by clinicians and researchers

- 2015 – Challenges in the care of familial hypercholesterolemia: a community care perspective (Brett, T., Watts, G.F., Arnold-Reed, et al.)
  - Scope: Australia
  - Findings: Community-based methods improve establishment of an index for contact tracing and cascade testing

- 2018 – Health Literacy in Familial Hypercholesterolemia: A Cross-National Study (Hagger, M.S., Hardcastle, S.J., Hu, et al.)
  - Scope: Global (AU, BR, CN, HK, MY, TW, UK)
  - Findings: Lower income and education are linked to inadequate health literacy, leading to access barriers, lower treatment adherence, and reduced participation in cascade screening

- 2020 – Women Living with Familial Hypercholesterolemia: Challenges and Considerations Surrounding their Care (Balla, S., Ekpo, E.P., Wilemon, K.A., Knowles, J.W. and Rodriguez, F.)
  - Scope: United States, United Kingdom
  - Findings: Women with FH face disparities: less likely to be on statins, more likely to discontinue treatment, and less likely to achieve LDL-C targets

- 2021 – Addressing Gaps in Racial/Ethnic Representation in Familial Hypercholesterolemia Registries (Mszar, R., Santos, R.D. and Nasir, K.)
  - Scope: United States
  - Findings: FH registries lack diversity; improvements can be made via population thresholds, revising diagnostic criteria, and engaging underserved communities with targeted research and interventions

- 2021 – Racial Disparities in Modifiable Risk Factors and Statin Usage in Black Patients With Familial Hypercholesterolemia (Agarwala, A., Bekele, N., Deych, E., et al.)
  - Scope: United States
  - Findings: Black FH patients have higher cardiovascular risk burdens and are less likely to receive lipid-lowering therapy, highlighting a need for targeted prevention

- 2022 – Familial Hypercholesterolemia Prevalence Among Ethnicities—Systematic Review and Meta-Analysis (Toft-Nielsen, F., Emanuelsson, F. and Benn, M.)
  - Scope: Global (BR, CN, DK, GE, JP, KR, MY, SA, US)

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  - Findings: High FH prevalence among Black populations, influenced by bias in screening, access barriers, and underrepresentation in clinical data; suggests increased targeted screening
- 2022 – **Using Healthcare Claims Data and Machine Learning to Identify Health Disparities for Individuals With Diagnosed and Undiagnosed Familial Hypercholesterolemia** (Ahmad, Z., Xing, C., Khera, A., et al.)
  - Scope: United States
  - Findings: Lipid-lowering therapy more often prescribed to white, high-income, and highly educated FH patients, revealing clear disparities

- 2023 – **LDL cholesterol targets rarely achieved in familial hypercholesterolemia patients: A sex and gender-specific analysis** (Schreuder, M.M., Hamkour, S., Siegers, K.E., et al.)
  - Scope: The Netherlands and Norway
  - Findings: Women with FH are less likely than men to reach LDL-C targets and to be on high-intensity statins.

- 2024 – **Familial hypercholesterolaemia in children and adolescents from 48 countries: a cross-sectional study** (Dharmayat, K.I., Vallejo-Vaz, A.J., Stevens, C.A., et al.)
  - Scope: Global
  - Findings: Genetic diagnosis and likelihood of being on lipid-lowering medication are lower in non-high-income countries; cardiovascular risk factors and disease rates are higher

- 2024 – **Sex Differences in Diagnosis, Treatment, and Cardiovascular Outcomes in Homozygous Familial Hypercholesterolemia** (Mulder, J.W., Tromp, T.R., Al-Khnifsawi, M., et al.)
  - Scope: Global
  - Findings: No large sex disparities in HoFH; men have higher incidence of myocardial infarction and coronary interventions

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PERFECTO stands for "Preventing the Preventable – Familial Hypercholesterolaemia Paediatric Screening for Cardiovascular Health." This project is focused on childhood screening programs for inherited high cholesterol, which runs in families. These programs help detect this serious cardiovascular disease (CVD) risk factor early, which can prevent premature heart attacks and other serious health complications. By implementing simple healthy living habits such as a healthy, low-fat diet, physical activity, and avoiding smoking, FH can be managed day-to-day alongside medication, promoting overall cardiovascular health (CVH).

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