FH Europe Foundation

FHEF Response:

EU Call for Evidence on Health Checks for Cardiovascular Diseases

FHEF Evidence Response to the Call for Evidence on the Council Recommendation on Cardiovascular Health Checks

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Part I. Background

1. Coordinated EU-level Action on Cardiovascular Health Checks

Cardiovascular diseases (CVD) remain the leading cause of death and disability in the European Union, affecting 62 million people and causing 1.7 million deaths annually[1]. The economic burden exceeds €282 billion per year, placing major pressure on healthcare systems, economies, and societies across Europe.[1] Despite advances in treatment, many cardiovascular conditions continue to be diagnosed too late, often only after irreversible damage or major cardiovascular events such as heart attacks or strokes have already occurred.

Recognising the growing burden of cardiovascular disease, the European Commission's 2025 EU Cardiovascular Health Plan: the Safe Hearts Plan[2] identifies prevention, early detection and screening as strategic priorities for reducing premature cardiovascular mortality by 25% by 2035.[4] The forthcoming Council Recommendation on cardiovascular health checks therefore represents an important opportunity to support Member States in implementing coordinated, evidence-based and equitable approaches to cardiovascular prevention.

Current screening and prevention efforts remain fragmented across Europe. Less than half of EU Member States have structured cardiovascular screening programmes in place, while significant inequalities persist between and within countries. Socioeconomically disadvantaged populations, children, women, rural communities, vulnerable groups, and ethnic minorities continue to face barriers to timely diagnosis and preventive care.[3,4] In addition, many inherited cardiovascular risk factors, including the very common forms like familial hypercholesterolaemia (FH) and elevated lipoprotein(a) [Lp(a)], and the rare forms of familial hyperlipidaemias like Homozygous FH and familial chylomicronaemia syndrome (FCS), remain critically underdiagnosed despite being detectable decades before clinical disease develops.

A coordinated EU-level approach can help address these shared challenges by promoting common prevention principles, supporting exchange of best practices, strengthening health data interoperability, and improving early identification of individuals and families at increased cardiovascular risk. Integrating life-course prevention, family-based screening, and personalised cardiovascular risk assessment into health checks would support more effective, sustainable, and equitable cardiovascular prevention across the European Union.

While implementation of cardiovascular health checks may require initial investments in workforce capacity, laboratory services, training, and digital infrastructure, it is expected these costs will be offset over time through reduced disease burden, lower healthcare expenditure, and improved productivity.[5]

FH Europe Foundation *welcomes this initiative and the strong focus within the Safe Hearts Plan on prevention, personalised medicine, digital innovation, AI, and reducing inequalities, recognising the importance of* ***integrated and life-course personalised prevention approaches, starting early in life, with a particular consideration for children and young people****.*

2. Familial Hyperlipidaemias – Preventable CVD Risk Factor

Approximately one in five people, an estimated 90 million EU inhabitants, live with one or more forms of familial hyperlipidaemias that expose them to an early, predictable yet preventable cardiovascular risk.[6]

Familial hyperlipidaemias, a collective term describing four inherited lipid disorders, include familial hypercholesterolaemia (FH), in both its heterozygous (HeFH) and homozygous (HoFH) forms, elevated lipoprotein(a) [Lp(a)] and familial chylomicronaemia syndrome (FCS). HeFH and elevated Lp(a) are

among the most common inherited drivers of premature CVD, while HoFH and FCS are rare and severe conditions associated with very high cardiovascular and metabolic risk.[6-10] These conditions are present from birth and run in families, but many individuals remain undiagnosed until a major cardiovascular event. For severe conditions such as HoFH and FCS, delayed diagnosis can lead to life-threatening complications already during childhood or adolescence.

Unlike many cardiovascular risk factors, inherited lipid disorders can be identified decades before clinical disease develops, creating a major opportunity for predictive and personalised prevention across the life course.[6-12] When screened for and diagnosed early and appropriately managed, CVD can often be prevented. Detailed information on each disorder, including description, prevalence, detection rate, disease burden, diagnosis, prevention and management, is provided in *Annex 1.*

FH Europe Foundation (FHEF) is the leading international network of patient organisations and individuals dedicated to improving the lives of people living with **familial hyperlipidaemias**, bringing together over 40 patient organisations and individual ambassadors, patients, and families from across more than 30 countries spanning the EU, wider Europe, Asia, Australia, the Middle East, and the Americas.[13] Combining patient advocacy, lived experience, public health expertise, and implementation knowledge, FHEF provides real-world insight into early screening, personalised prevention, and inequalities in diagnosis and access to care across different healthcare systems.

The scientific, clinical, economic, and policy foundations for action in the area of familial hyperlipidaemias are already well established. Relevant clinical research and medical guidelines consistently demonstrate that early detection and management of FH (HeFH and HoFH), elevated Lp(a), and FCS significantly reduce cardiovascular risk and prevent avoidable events.[14-16] Economic evidence shows that childhood detection and family-based cascade screening are cost-effective and reduce long-term healthcare costs, disability, and productivity loss, while lived experience highlights the consequences of delayed diagnosis, fragmented care pathways, and the intergenerational impact of undetected inherited risk. [17,18]

The policy case for action is equally strong. The *Safe Hearts Plan*, the Council Conclusions on Cardiovascular Health (2024), as well as the Prague Declaration of FH Paediatric Screening and Brussels International Declaration on Lp(a) testing and management, all prioritise prevention, early detection, personalised medicine, and reduction of health inequalities.[19-21]

Furthermore, Joint Actions such as JACARDI demonstrate the need and opportunity to build stronger synergies across disease prevention initiatives.[22] Recognising the shared potential for early childhood prevention, the familial hyperlipidaemias and diabetes communities came together around conditions that have a similar prevalence in Europe, begin early in life, and can be identified through simple blood-based screening tests before serious complications develop. This creates important opportunities for coordinated paediatric screening pathways linking inherited lipid disorders and type 1 Diabetes (T1D).

The forthcoming Council Recommendation should therefore ensure that inherited lipid disorders become a core component of EU cardiovascular health checks across the life course. This should include paediatric screening, early detection, accurate diagnosis, and paediatric screening pathways linked to T1D initiatives, preferably in the first decade of life. Such an approach would represent a highly implementable and cost-effective step towards reducing avoidable CVD, improving health equity, and strengthening preventive healthcare across Europe.

3. The Continued Public Health Gap

Inherited cardiovascular risk remains insufficiently recognised within public health strategies and routine prevention programmes. FH and elevated Lp(a) are among the most important inherited drivers of premature CVD, yet detection rates remain critically low across Europe. The estimated rate for FH is 10-15% diagnosed, for elevated Lp(a) of 2% diagnosed, and no sufficient data for FCS are available.[7,21] As a result, millions of individuals and families continue to live with predictable and preventable high cardiovascular risk without knowing it.

Significant fragmentation also persists across Member States. Approaches to lipids in particular correct FH screening, diagnosis, reimbursement, registries, family-based cascade screening and equitable access to specialised care vary considerably between countries and even within healthcare systems. While some Member States have introduced structured screening initiatives or national FH programmes, many still lack coordinated pathways for early detection and long-term management of inherited lipid disorders.

> *"Early diagnosis should not depend on tragedy, and it should not depend on luck. When my father suffered a heart attack at 30 and was diagnosed with FH, it set off a search for answers. Tests revealed that I also had FH. I was 2 years old. Unlike my father, my journey began with prevention, not crisis. That difference, early versus late detection, separated survival from tragedy. Today, I am studying genetics, determined to ensure that no family must repeat my story. My experience embodies the message that prevention must begin early and generationally."*
>
> **Lena-Rosa Hanauer, 25, Austria**

Inequities in access to diagnosis and care further widen this gap. Socioeconomic status, geography, healthcare literacy, and access to specialised services continue to influence whether individuals receive timely diagnosis and treatment.[3] Important disparities also exist in access to: Lp(a) testing, where evidence has been found demonstrating that such testing, despite being available and reimbursed, is actively not offered; genetic testing as a confirmatory test for FH (HeFH, HoFH) and FCS; specialist referral and preventive therapies.

Women and children with FH remain particularly underdiagnosed and underserved. Cardiovascular risk in women is still frequently underestimated, while sex-specific risk factors and the impact of pregnancy-related complications are often insufficiently integrated into cardiovascular prevention strategies.[4] At the same time, many children living with inherited lipid disorders remain unidentified despite the fact that these conditions are present from birth and early intervention can substantially reduce lifetime cardiovascular risk.[7,20]

> *I was diagnosed with familial hypercholesterolaemia earlier this year (2025). […] Luckily, my diagnosis came early enough in life for me to be able to manage this disorder in a way that will allow me a regular, healthy life. My father wasn't as fortunate. He was only diagnosed with FH after his first heart attack had already happened. By then, irreversible damage had occurred. He went on to develop serious cardiovascular and atherosclerotic disease and, ultimately, passed away at 63 from complications that might have been prevented with early detection and proper care. Our story is not unique. […] The EU now has the opportunity to change the future for millions of families like mine.*
>
> **Madalina Iamandei, 37, Romania**

Current approaches, therefore, continue to prioritise treatment of advanced disease rather than prevention of disease development. This is neither sustainable nor aligned with the objectives of the EU Safe Hearts Plan. Europe now has an opportunity to shift towards a more predictive, preventive and life-course approach to cardiovascular health—one that identifies inherited risk earlier, integrates family-based prevention and ensures equitable access to screening, diagnosis and management across all Member States.

I was a healthy father of three. I was fit, did a lot of sport and never smoked. Only to experience a heart attack at the age of 39. "Bad luck"- I was told. It took 6 very long years and two heart attacks, until diagnosis and the root cause of my heart/artery issues was identified. As elevated levels of Lp(a) is genetic, my family was also tested and two of my daughters unfortunately inherited this disorder. This was difficult to accept at first, but currently I am confident that they will be able to live a healthy life (now that they are aware).

Marc Rijken, 50, The Netherlands

4. Effective Cardiovascular Health Checks Across the Life Course

For the forthcoming Council Recommendation on cardiovascular health checks to deliver meaningfully on the Safe Hearts Plan objective of reducing premature cardiovascular mortality by 25% by 2035, cardiovascular prevention must move beyond a predominantly reactive, adult-focused model towards a predictive, personalised, life-course and family-based approach to prevention.[2]

While current approaches largely focus on identifying cardiovascular risk in adulthood, many of the strongest drivers of premature CVD are already present at birth. Familial hyperlipidaemias, including FH, in both forms HeFH and HoFH, elevated Lp(a), and FCS, represent among the most common inherited causes of premature CVD, yet remain critically underdiagnosed across Europe despite the availability of simple, low-cost, and clinically actionable screening tools.[6-12,17,18]

Based on over a decade of patient-led evidence generation, implementation experience, EU-funded research projects, clinical collaboration, and international consensus-building with the leading societies, including the European Atherosclerosis Society (EAS)[22], the International Atherosclerosis Society (IAS)[24], and other partners like the diabetes community, FHEF strongly recommends that the Council Recommendation explicitly include:

- systematic measurement of a full lipid profile, including Lp(a);
- early paediatric lipid screening within the first decade of life;
- universal and opportunistic screening approaches adapted to national healthcare systems;
- family-based reverse and cascade screening pathways;
- integration of inherited lipid disorders into cardiovascular risk prediction and prevention strategies; and
- coordinated paediatric screening pathways linking inherited lipid disorders with T1D early detection initiatives.

"I was diagnosed as a child at the age of 6, as part of the routine family cascade screening in the Netherlands. My dad had a family history of high cholesterols and CVD, and then we heard of FH. To everyone's surprise I didn't have FH, but elevated Lp(a). This triggered the reverse cascade screening. And this is how my mom was diagnosed with elevated Lp(a). The family-based screening has unraveled a complex genetic mix in our family and at the same time helped identify everyone at risk early enough to prevent any tragedy."

Aeden Kaal, 19, The Netherlands

5. Detecting Risk Before Disease: Screening

Strong scientific, clinical, and economic evidence demonstrates that early identification of inherited cardiovascular risk factors combined with family-based prevention is effective, feasible, and cost-effective.[16,17] Evidence from multiple European countries shows that paediatric screening linked to reverse and cascade testing enables earlier identification of at-risk families and prevention of avoidable cardiovascular events[11].

Rather than waiting for the first heart attack or stroke, early detection reframes people not as patients defined by disease, but as individuals with manageable risk factors who can benefit from timely, preventive management. Systematic early screening for inherited cardiovascular risk factors, including FH and elevated Lp(a), is essential to shift from reactive care towards preservation of cardiovascular health (CVH) in line with the Safe Hearts Plan. The Council Recommendation should support a structured life-course approach to cardiovascular prevention built on childhood detection, family-based prevention and adult cardiovascular risk assessment.

5.1. A childhood health check for familial hyperlipidaemias and Type 1 Diabetes

Recognising the shared opportunity for early childhood prevention, the inherited lipid disorders and the diabetes communities came together around FH and T1D, which share the same estimated prevalence, begin early in life, and can be identified through simple blood-based screening before serious complications develop. A dedicated multidisciplinary Task Force was therefore established bringing together the FHEF[13], the EAS[23], the Lp(a) International Taskforce (Lp(a) ITF)[25], Breakthrough T1D[26], the European Diabetes Forum (EDF)[27], International Diabetes Federation Europe (IDF Europe)[28] and International Society for Paediatric and Adult Diabetes (ISPAD)[29], with the endorsement from the consortia and organisations PERFECTO FH[30], FH-EARLY[31], PerMed FH[32], European Association for the Study of Diabetes (EASD)[33], EDENT1FI[34], Foundation of European Nurses in Diabetes (FEND)[35], Société Francophone du Diabète[36], and Global Heart Hub (GHH)[37]. The Task Force developed a joint early childhood screening protocol integrating inherited lipid disorders and T1D into coordinated prevention pathways aligned with the objectives of the Safe Hearts Plan.

*The Task Force on defining a **childhood health check for inherited lipid disorders and type 1 diabetes** invites the Commission to integrate the proposed structured approach for rolling out childhood health checks across the 27 EU Member States and to reflect the following wording in the upcoming Council Recommendation as part of the European Safe Hearts Plan.*

Rationale: Inherited lipid disorders such as familial hypercholesterolaemia (FH), elevated lipoprotein(a) [Lp(a)], familial chylomicronaemia syndrome (FCS), and type 1 diabetes are early-onset conditions with identifiable preclinical phases, validated biomarkers, and clear clinical benefits from early identification and intervention. Collectively, they affect an estimated 95 million people across 27 Member States.

Inherited lipid disorders can be detected in children by the measurement of **a full lipid profile [total cholesterol, low-density lipoprotein (LDL) cholesterol, high-density lipoprotein (HDL) cholesterol, triglycerides, and lipoprotein (a) (Lp(a))]**.

Type 1 diabetes can be detected in children in early stages, prior to the insulin-requiring clinical diagnosis, through screening for **islet auto-antibodies against insulin, IA2, ZnT8, and GAD.**[38]

Combining screening for inherited lipid disorders and type 1 diabetes in childhood would create efficiencies in delivery, improve uptake, and enhance prevention of both acute metabolic complications and long-term cardiovascular (CV) risk. A combined approach reflects the Safe Hearts Plan's emphasis on early detection,

lifelong prevention, and coordinated care pathways. Existing evidence: scientific, medical, economic, behavioural, and related to quality of life and patient preferences, strongly supports the early screening approach to detect inherited lipid disorders and type 1 diabetes, and effectively prevent CVD across families and generations.

Screening: Systematic lipid and type 1 diabetes screening for all children in the first decade of life is recommended to detect children with early-stage inherited lipid disorders and type 1 diabetes by measurement of a **full lipid profile and through screening for islet auto-antibodies.**

For any screening, the age ranges indicated can be adapted to the screening opportunities provided within established public health activities.

Integrated with childhood health checks, Member States should also promote awareness, genetic counselling, family-based cascade screening, and implementation of personalised and guideline-based management pathways for treating identified conditions and reducing CV risk.

5.2. Family-based Screening

Furthermore, adopting family-based approaches (cascade and reverse cascade screening) creates cost savings as one diagnosis triggers cascade or reverse-cascade testing to protect whole families across generations.

Cascade screening is a method used to identify individuals within a family who may be at risk of inheriting specific genetic conditions. It involves screening close relatives of a person diagnosed with a specific genetic condition to determine if they also have the same genetic mutation.

In the case of FH and elevated Lp(a), early intervention can help manage LDL (“bad”) cholesterol levels and reduce the risk of cardiovascular disease. Note, *Lp(a) testing does not require genetic testing.*

This approach, when implemented correctly, allows families and health systems to act decades ahead of irreversible damage, turning a reactive model into one that promotes resilience, longevity, and improved quality of life.

5.3. Global Cardiovascular Risk Assessment

Global cardiovascular risk assessment in adults is essential to shift from late-stage disease management towards earlier identification and prevention of cardiovascular and metabolic risk. Health checks should begin sufficiently early in adulthood to avoid missing decades of prevention opportunities and to enable timely intervention before irreversible cardiovascular damage develops.

Systematic health checks are recommended, and a basic one should be performed at the age of 18-20 years. The aim is early detection of risk factors and behaviours, promotion of self-awareness, education, behaviour change, and initiation of treatment if indicated. If no abnormalities are found, it should be repeated every 5 years for all adults aged 35 to 65 years. These basic and repeated health checks should focus on early detection and timely management of key cardio-renal-metabolic (CRM). A complete assessment comprises all of the following elements:

First, evaluation of lifestyle and health behaviours, including diet, physical activity, sedentarism, smoking or vaping, alcohol and other toxins, body mass index (BMI), and waist circumference or waist-to-height ratio to assess overweight and central adiposity; and assessment of mental health disorders, such as depression or anxiety, using abbreviated (2-item) questionnaires.

Second, measurement of traditional cardiovascular risk factors, including blood pressure, full lipid profile (total cholesterol, LDL-C, HDL-C, triglycerides), and Lp(a), the latter at least once in adulthood.

For assessment of glucose metabolism, fasting plasma glucose (FPG) and glycated haemoglobin (HbA1c) should be used to detect diabetes and pre-diabetes;

Measurement of kidney function is calculated through estimated glomerular filtration rate (eGFR) from serum creatinine (using the CKD-EPI formula) and albuminuria, typically measured as Urinary Albumin:Creatinine Ratio (uACR).

For individuals over 65 years, a full metabolic check and basic cardiovascular assessment should be performed not less than every 3-5 years, including natriuretic peptide tests (NT proBNP) for the early detection of heart failure, pulse (heart rate and rhythm), cardiac auscultation, and evaluation of peripheral pulses for peripheral artery disease (PAD). The objectives are to monitor control of known risk factors, identify new-onset factors, detect previously unrecognised organ damage, and facilitate the rule-out of subclinical cardiovascular disease.

Part II. The Evidence Base

6. Evidence Generation

This submission builds on more than a quarter century of scientific evidence, policy development, and international collaboration on inherited lipid disorders, beginning with the World Health Organization (WHO) reports on FH published in 1997 and 1998, followed by the 2015 *Call to Action on Familial Hypercholesterolaemia (FH) and Cardiovascular Diseases* and the 2020 Global Call to Action on FH.[39-42] It also reflects over a decade of community building, lived experience engagement, and evidence generation led by FH Europe Foundation (FHEF) together with patients, families, clinicians, researchers, and policymakers across Europe and beyond.

Through continuous collaboration with patient organisations, ambassadors and advocates, FHEF has gathered real-world insight into the impact of delayed diagnosis, fragmented care pathways and inequalities in access to screening, diagnosis and treatment for inherited lipid disorders.

The central focus of this work has been understanding and comparing experiences of early screening and detection across different healthcare systems, particularly in relation to FH, elevated Lp(a), HoFH and FCS. These lived experience insights have consistently highlighted the need for earlier identification, family-based prevention, and more coordinated cardiovascular screening pathways across Europe.

This work contributed to the recognition of FH Paediatric Screening as a European Best Practice on the EU Public Health Best Practice Portal in 2021, building on implementation experience from the Netherlands, Slovenia, Czechia, and Austria, and supporting broader collaboration with patient organisations, scientific societies, and public health stakeholders across multiple European initiatives.[42]

FHEF's evidence generation activities were further strengthened through high-level policy and scientific discussions organised under the Slovenian (2021), Czech (2022), and Polish (2025) Presidencies of the Council of the European Union, as well as consensus-building initiatives involving patients, clinicians, researchers, policymakers, politicians, and public health experts.[43-46] These efforts resulted in important policy and scientific outputs, including the *Prague Declaration on FH Paediatric Screening* (2022) and the *Brussels International Declaration on Lp(a) Testing and Management* (2025), helping establish broad expert consensus on early paediatric FH screening combined with family-based reverse and cascade screening, life-course prevention, and equitable access to diagnosis and care for inherited lipid disorders.[20,21]

As part of the Lp(a) International Taskforce, FHEF also commissioned the first independent cost-effectiveness study on Lp(a) testing in primary prevention, strengthening the economic case for population-based and risk-based screening approaches (2024).[18,25] The findings were widely disseminated and discussed during several European Atherosclerosis Society (EAS) Congresses and other international scientific meetings.

The submission is further supported by findings from major research and implementation projects, including:

- *PERFECTO FH* (EU4Health),[30]
- *FH-EARLY* (Horizon Europe),[31] and
- *PerMed FH* (Fundación "la Caixa").[32]

Together, these projects generated evidence on:

- early screening and detection pathways;
- personalised prevention strategies;
- patient engagement and health literacy;
- inequalities in diagnosis and care; and
- implementation of family-based cardiovascular prevention across healthcare systems.

FHEF also organised the high-level European Parliament event *Cardiovascular Prevention as the Cornerstone of a Competitive Europe: Scaling Up Lipid Screening to Secure Next Generations* (2025), hosted by Members of the European Parliament Romana Jerković and Tomislav Sokol.[46] The event reinforced the growing political and scientific consensus that early detection of inherited lipid disorders should become a strategic component of European cardiovascular prevention policy.

Most recently, FHEF co-led a dedicated European Alliance for Cardiovascular Health (EACH) Task Force on cardiovascular health checks, which successfully developed and secured adoption of a joint health check protocol by all EACH partners (2026).[48] The collaboration also included close engagement with the diabetes community, resulting in the development of a dedicated paediatric screening protocol linking inherited lipid disorder screening with T1D prevention pathways.

The 2026 closing event of *PERFECTO FH*[49] provided an opportunity to present additional evidence on equity in FH paediatric screening, including examples of 12 national screening programmes, pilots, and implementation initiatives across Europe, among them the Latvian pilot developed under JACARDI[22].

In preparing this submission, FHEF consulted its international network of patient organisations, advocates, and scientific experts through dedicated community consultations and structured feedback processes, ensuring that the recommendations presented reflect both scientific evidence and lived experience across diverse healthcare systems.

7. Registry, Consensus Statements, Recommendations, and Guidelines

The submission is further supported by a substantial body of existing data from the EAS FH Study Collaboration, the global registry (EAS FHSC)[50], the Homozygous Familial Hypercholesterolemia International Clinical Collaboration (HICC)[51], the international scientific consensus statements and expert recommendations and subsequently clinical guidelines, recognising inherited lipid disorders as major drivers of premature CVD and supporting earlier detection, family-based screening and life-course prevention approaches. Key reference documents include:

- European Atherosclerosis Society (EAS) Consensus Statements on FH, HoFH, paediatric FH, FCS and Lp(a)[6,14,15,52];
- European Society of Cardiology (ESC) and European Atherosclerosis Society (EAS) Guidelines on CVD prevention and dyslipidaemias[14,15];
- the Prague Declaration on FH Paediatric Screening[20];
- the Brussels International Declaration on Lp(a) Testing and Management[21];
- the International Atherosclerosis Society (IAS) global recommendations on FH[53,54]; and,
- World Health Organization (WHO) reports and recommendations on FH[39,40]

Together, these guidelines and consensus documents provide a strong scientific and clinical foundation for integrating inherited lipid disorders into EU cardiovascular health checks and broader prevention strategies.

8. Cost-Effectiveness of FH Screening & Lp(a) Testing

CVD prevention strategies that identify high-risk individuals early, particularly those with FH or elevated Lp(a), can avert premature events and long-term treatment costs. Economic evaluations consistently show that well-designed screening programmes for these conditions deliver good value for money in high-income settings, especially when paired with effective treatment pathways and cascade testing of relatives. Evidence from studies on FH, including country-specific and systematic reviews and elevated Lp(a) demonstrates that early screening and testing are not only cost-effective but also cost-saving, delivering measurable benefits for both healthcare systems and society as a whole.[17,18,53]

8.1. Country example – FH Cascade Screening

Evidence from the Netherlands demonstrates both health and economic benefits of early FH detection.[17] A 20-year national cascade screening programme identified over 26,000 FH carriers, including 5,600 children, by systematically tracing families from index cases. Health economic modelling of paediatric screening showed that early detection and statin initiation at age 10 yields an average gain of 2.53 quality-adjusted life years gained per person compared to late diagnosis, at an incremental cost-effectiveness ratio of only €9,220 per QALY—well below the Dutch threshold of €20,000. From a broader societal perspective, the programme was cost-saving, with **every €1 euro invested generating an estimated €8.37 return on investment through reduced premature heart attacks, hospitalisations, and productivity losses**. These findings confirm that systematic paediatric FH screening is not only life-saving but also delivers excellent value for European health systems.

Once-in-a-lifetime Lp(a) testing in adults, for example, at the first comprehensive cardiovascular disease (CVD) risk assessment, is generally cost-effective in high-income health systems as a risk-refinement tool, especially for individuals at borderline to intermediate risk or with a family or personal history suggestive of elevated Lp(a).[18] As Lp(a) is genetically determined and stable throughout life, a single test provides a lifelong signal for cardiovascular risk management. Testing enables risk reclassification by identifying a subset of individuals at substantially higher risk, supporting intensified prevention strategies such as stricter LDL-C lowering targets, aspirin in select secondary prevention contexts according to guidelines, and earlier imaging where appropriate. It also improves downstream efficiency by focusing healthcare resources, including clinics, imaging, and pharmacotherapy, on individuals with the greatest expected benefit.

8.2. Regional example – Lp(a) Testing

A recent multinational health-economic analysis in 11 high-income countries, including France, Germany, Italy, the Netherlands, Spain, Poland, Austria and the UK, demonstrated that universal Lp(a) testing in primary prevention is cost saving (more effective and less costly).[18] Health-economic modelling demonstrated that testing would prevent 60 first heart attacks, 13 strokes and 26 premature deaths per 10,000 individuals, and would lower healthcare costs between ≈75-450€ per person in the explored EU countries. Reclassification of almost 20% of adults into higher-risk categories occurred only when Lp(a) was added jointly with traditional cardiovascular risk factors. This led to earlier initiation of intensive LDL-C lowering and better blood pressure control, helping to prevent heart attacks and strokes while avoiding costly revascularisations. From a societal perspective, incorporating productivity losses due to premature mortality, workforce dropouts, absenteeism, and presenteeism, Lp(a) testing intervention was cost-saving from both the healthcare and societal perspectives as supported by the analysis across European health systems.

Part III. Equity and Social Justice

9. FH, Women and the Need for Earlier Detection

FH affects women and men equally, yet women remain systematically underdiagnosed, undertreated, and less likely to achieve guideline-recommended LDL-cholesterol targets. Although FH affects approximately 1 in 300 people globally and causes lifelong exposure to elevated LDL-cholesterol from early childhood, the average age of diagnosis remains around 44 years, with only about 2% of cases diagnosed in childhood and 17% already having CVD at diagnosis.[7]

This late detection has specific consequences for women. Evidence shows that women with FH are typically diagnosed 3 to 7 years later than men, are 26% less likely to receive lipid-lowering therapy, and are 37% less likely to achieve recommended LDL-cholesterol targets.[55] These gaps reflect broader sex and gender inequities in cardiovascular prevention, where women's cardiovascular risk is often underestimated, and treatment is delayed or less intensive.

Women with FH also face unique challenges during reproductive years. Lipid-lowering therapy, particularly statins, is generally interrupted from pre-conception, throughout pregnancy, and often until the end of breastfeeding, resulting in prolonged periods without treatment during key stages of life. Recent evidence demonstrates that the cumulative pregnancy-related off-treatment time after childbirth is a median of 2.9 years per woman, ranging from 0.8 to 12 years depending on the number of pregnancies and breastfeeding duration. Importantly, pregnancy itself accounted for only around 42% of this off-treatment period, while the majority of treatment interruption occurred before conception and after pregnancy during breastfeeding and family planning phases.[55]

When untreated childhood years and delayed diagnosis are also considered, women with FH spend a median of 66.3% of their lifetime without lipid-lowering treatment, with some women remaining untreated for virtually their entire lives. This creates a major cumulative cholesterol burden during precisely the years when cardiovascular prevention could have the greatest long-term impact.[55,56]

These realities create a particularly strong public health and clinical rationale for earlier detection of FH in girls and young women. Identifying FH early in life allows preventive treatment to begin years before reproductive age, helping compensate for future unavoidable treatment interruptions associated with pregnancy and breastfeeding. Earlier initiation of treatment substantially reduces cumulative LDL-cholesterol exposure and may delay or prevent the onset of atherosclerotic cardiovascular disease (ASCVD) before cardiovascular damage develops. Early diagnosis also enables appropriate counselling, personalised cardiovascular risk management, and family-based prevention strategies long before pregnancy planning occurs.

The forthcoming Council Recommendation should therefore explicitly recognise FH screening and early detection in girls and women as an essential component of cardiovascular prevention and women's health. This should include paediatric and adolescent screening, systematic family history assessment, earlier identification of inherited cardiovascular risk, pre-conception counselling, pregnancy-aware lipid management pathways, and structured follow-up after pregnancy and breastfeeding. Detecting FH early is not only a paediatric prevention measure; it is also a critical women's cardiovascular health intervention that can reduce lifelong cardiovascular risk and prevent avoidable premature ASCVD across generations.

10. FH Paediatric Screening and Health Equity

Health equity is a critical consideration in FH paediatric screening. Social, economic, and structural barriers continue to limit access to early diagnosis, screening, and care for inherited lipid disorders, particularly among underserved populations, including migrants, Roma communities, and socioeconomically disadvantaged groups.[57-68]

Using the Dahlgren-Whitehead model on social determinants of health, evidence shows that disparities in FH are shaped not only by biology, but also by healthcare access, health literacy, socioeconomic conditions, and trust in healthcare systems.

Key inequities include[57-68]:

- unequal access to diagnosis, genetic testing, and treatment;
- lower participation in family cascade screening;
- under-representation of vulnerable populations in research and registries;
- geographical and income-based disparities in specialist care;
- lower health literacy and culturally inappropriate communication; and
- persistent gender inequalities in diagnosis and treatment.

Addressing these gaps requires more equitable, community-based, and culturally adapted screening approaches integrated into broader public health and primary care systems.

Core Recommendations for EU Cardiovascular Health Checks

The forthcoming Council Recommendation should support a coordinated life-course approach to cardiovascular prevention and explicitly include:

- systematic measurement of a full lipid profile, including lipoprotein(a) [Lp(a)];
- early paediatric lipid screening within the first decade of life;
- universal, targeted, and opportunistic screening approaches adapted to national healthcare systems and population needs;
- family-based reverse and cascade screening pathways;
- coordinated paediatric screening pathways linking inherited lipid disorders with Type 1 Diabetes (T1D) early detection initiatives;
- integration of inherited lipid disorders into global cardiovascular risk assessment, prediction, and prevention strategies across adulthood, reflecting interconnected cardiovascular, metabolic, kidney, and mental health risk factors across the life course;
- systematic consideration of family history of premature cardiovascular disease to improve identification of inherited cardiovascular risk;
- equitable implementation approaches ensuring access to early detection, diagnosis, prevention, and care for all individuals and families affected by inherited lipid disorders, including underserved populations, women, children, minorities, rural populations, and those living with rare conditions such as homozygous familial hypercholesterolaemia (HoFH) and familial chylomicronaemia syndrome (FCS);
- development and responsible use of interoperable digital health tools, electronic health record (EHR)-based screening algorithms, registries, and AI-assisted cardiovascular risk assessment systems aligned with the European Health Data Space (EHDS);

Furthermore, the Council Recommendation should support interoperable registries, secondary use of health data, and structured involvement of patient organisations, citizens, cities, and communities in the co-design and implementation of cardiovascular health checks. It should promote dedicated support for newly detected patients and families through health literacy, peer support, culturally adapted communication, and personalised communication models (PCM), particularly in underserved populations. Continuous monitoring, patient-reported outcomes, and social return on investment (sROI) approaches should help ensure equitable, sustainable, and person-centred implementation aligned with the Safe Hearts Plan, the European Health Union, and the European Health Data Space.

Annex 1. Familial Hyperlipidaemias

Familial hypercholesterolaemia (FH) is an umbrella term for common and rare genetic high cholesterol, leading to lifelong high LDL-cholesterol and a markedly increased risk of premature coronary heart disease, running in families.[7,8]

FH is one of the most common inherited cardiovascular risk factors, yet one of the least recognised. It has a more common form, called heterozygous FH (HeFH), and a rarer, more severe form, called homozygous FH (HoFH). With HeFH (inherited from one parent), cholesterol levels more than double, and heart attacks can strike decades too early. With HoFH (inherited from both parents), cholesterol is extremely high, and heart attacks can occur already in childhood.[8]

FH literally means "running in the family (inherited from parents, passed to children), very high cholesterol in the blood, from birth onwards."

When one parent has FH and the other parent does not, each child has:

- 50% probability of inheriting HeFH, when one FH-causing gene is inherited from the affected parent.
- 50% probability of inheriting unaffected genes and therefore not having FH.

When both parents have FH, the chances of passing it on change, resulting in the following possibilities:

- 50% probability of inheriting **HeFH**, when a single gene is mutated.
- 25% probability of inheriting **HoFH,** when two of the same FH-causing genes are mutated.
- 25% probability of inheriting two unaffected genes that don't cause FH.

FH can be diagnosed based on clinical criteria; however, if feasible, genetic testing for causative genes should be undertaken with testing for pathogenic variants in 3 primary genes**: LDLR, APOB, and PCSK9.**

Cascade screening allows for testing of family members of the index patient (the proband) who are at risk based on their relationship to the patient; this can be done through a combination of lipid profile screening and genetic testing. Among those with genetically confirmed FH, cascade screening represents a simple, cost-effective method for identifying additional cases in a family. This ultimately leads to earlier diagnoses of FH and promotion of personalised lipid-lowering therapy.[7,12]

"Think of it as being born with a ticking bomb in your arteries. From birth, bad cholesterol is dangerously high and rapidly builds up fatty deposits in the artery walls. FH is not lifestyle, it's DNA. But it can be detected with a simple test and treated effectively".

Prof. Albert Wiegman, paediatric cardiologists, UMC Amsterdam

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Heterozygous FH (HeFH)

Prevalence: 1 person in 300 has HeFH, an estimated 2.5 million Europeans, including 500,000 children.[7]

Detection rate: It is estimated that only 10-20% of people with HeFH have been diagnosed, and fewer than 5% of children have been identified.[7] Most cases are still detected following a heart attack in secondary prevention.

Burden: 22-fold increased risk of premature coronary heart disease compared with people with normal LDL-C and much higher than persons with high cholesterol without a genetic cause (6-fold). Untreated HeFH increases the risk of early heart attacks. Around 50% of men with HeFH experience a heart attack by age 50, and 30% of women with HeFH by age 60. Avoidable hospitalisations, productivity losses, higher sick leave, and early retirement often follow. The average age of the first heart attack in people with undiagnosed and untreated HeFH is 40.[7]

Diagnosis: Universal paediatric screening (age 5-10) and family cascade and reverse cascade testing identify cases early and meet all Wilson-Jungner criteria.[69]

Prevention and management: Early statin therapy can normalise life expectancy from childhood. Access to available lipid-lowering therapies, combined with lifestyle changes with smoking cessation, physical activity, and diet.[7]

Homozygous FH (HoFH)

Prevalence: One of the rarest and most severe inherited cholesterol disorders, it affects 1 in 250,000-360,000 people, about 10,000 citizens across Europe.[8]

Detection rate: fewer than 1 in 20 are diagnosed, and many are identified in their teenage years, when serious heart damage has already begun.[8]

Burden: Children with HoFH are born with extremely high cholesterol that damages arteries from the very start of life. Without specialist care, heart attacks can appear in early childhood, and fatal cases have been reported as young as 1.5 years of age. Most patients never reach safe cholesterol levels, facing a constant risk of early heart attacks, strokes, and premature death.[8,11]

Diagnosis: Very high cholesterol in blood, visible cholesterol deposits under the skin and around the eyes (xanthomas), family history of FH, high cholesterol, heart attacks, and premature death in family. Diagnosis should be confirmed through genetic testing. Implementation of cascade and reverse cascade testing of family members is critical to diagnose other family members due to the genetic nature of the disorder.[8,11]

Prevention and management: When HoFH is identified early, children can access modern, life-saving treatments, including specialised medicines and treatments that dramatically reduce risk. Linking these children to expert lipid clinics and testing their families (reverse cascade screening) is critical to protect lives across generations.[8,11]

Elevated Lp(a)

Description: Elevated Lp(a) is an inherited, independent, and causal risk factor for atherosclerotic cardiovascular disease (ASCVD), including coronary artery disease, stroke, peripheral arterial disease, and aortic valve stenosis. Around 90% of levels are genetically determined and remain stable from childhood, unaffected by lifestyle. Levels may vary slightly in women later in life due to hormonal changes.[6]

Prevalence: About 1 in 5 people, nearly 20% of Europeans, have elevated Lp(a) above the high-risk threshold of 50 mg/dL, and it is therefore the most common inherited lipid disorder in humans.[6]

Detection rate: Only 1-2% of citizens are tested, despite the availability of simple, low-cost assays and **no genetic testing is required**.[6,20]

Burden: Elevated Lp(a) can raise the risk of heart attack and stroke by 50–300%, and one Lp(a) particle is estimated to be six times more atherogenic than one LDL particle on a particle-for-particle basis.[6] Each year, elevated Lp(a) is responsible for thousands of preventable premature cardiovascular events, including early heart attacks, strokes, and cases of aortic valve stenosis. Importantly, Lp(a) should always be evaluated in the context of overall cardiovascular risk, taking into account other factors such as high cholesterol, diabetes, hypertension, and obesity, etc., since this strongly guides the management of high Lp(a).[6,20]

Diagnosis: A once-in-a-lifetime blood test is sufficient to identify elevated Lp(a), as the levels stabilise at the age of 5. Implementation of cascade and reverse testing of family members is critical to diagnose other family members due to the genetic nature of the disorder.[20] However, since the condition is so frequent, systematic testing of the general population is recommended by guidelines.[13,14]

Prevention and management: While specific Lp(a)-lowering treatments are still in development, risk can be reduced through intensive management of other cardiovascular risk factors: lowering LDL cholesterol as much as possible, controlling blood pressure, smoking cessation, maintaining healthy weight and diet, and increasing physical activity. Identifying elevated Lp(a) early enables clinicians to reclassify risk, adjust treatment plans, and protect families across generations.[20] These recommendations will still be valid after specific Lp(a)-lowering drugs become available, since these drugs will only be available for patients in the secondary prevention setting. For all other individuals affected by high Lp(a) the management of all traditional risk factors for CVD will be of utmost importance. Therefore, knowing the Lp(a) concentrations should have an overall influence on guiding the risk factor management of a given person.

Lp(a) Testing and Global Risk Management

The absence of an approved, specific Lp(a)-lowering therapy is not a sufficient reason to exclude Lp(a) testing from cardiovascular health checks. Elevated Lp(a) is a common, inherited, independent, and causal cardiovascular risk factor, affecting around one in five people, while current testing rates remain extremely low at only 1–2%. Lp(a) levels are largely genetically determined, stable across life, and run in families, meaning that one test can inform lifelong risk management and enable cascade testing of relatives.[6]

Testing does not diagnose CVD; it identifies a biologically determined risk pathway that can and should be managed in the context of global cardiovascular risk. The impact of elevated Lp(a) increases substantially when combined with other risk factors such as high LDL cholesterol, high blood pressure, diabetes, smoking, obesity, family history, and unhealthy lifestyle. The Brussels Declaration stresses that Lp(a) should never be assessed in isolation but used to reclassify cardiovascular risk and develop a personalised cardiovascular health roadmap.[20]

In the absence of specific Lp(a)-lowering medicines, the recommended response is early, intensive management of all modifiable cardiovascular risk factors, including LDL cholesterol, blood pressure, glucose, smoking, weight, diet, and physical activity. The 2022 EAS consensus statement explicitly recommends early risk factor management for people with elevated Lp(a), taking into account both absolute global cardiovascular risk and Lp(a) level.[6]

Early testing, including earlier in the life course, is therefore clinically and ethically justified. It prevents missed years of prevention, enables family-based risk identification, supports personalised prevention, and gives individuals and families the opportunity to act before irreversible cardiovascular damage occurs. For this reason, Lp(a) testing should be included in EU cardiovascular health checks as a once-in-a-lifetime test, ideally early in life and alongside LDL cholesterol, with results embedded in global cardiovascular risk assessment, family cascade testing, and equitable access to preventive care.[6,14,20]

Familial Chylomicronaemia Syndrome (FCS)

Description: FCS is an ultra-rare inherited lipid disorder causing extremely high triglycerides and recurrent, sometimes life-threatening pancreatitis. It requires lifelong management and specialist care.[9]

Prevalence: The best available estimates suggest that FCS affects approximately 1 in 1,000,000 people, with higher prevalence observed in certain founder populations.[9]

Detection rate: Due to the rarity of the disease, many patients are estimated to be undiagnosed or misdiagnosed for years.[9] More research would be needed to better assess the misdiagnosis rate.

Burden: FCS causes severe abdominal pain, recurrent hospital admissions for acute pancreatitis, and major psychosocial impacts. Patients often face restrictive, extremely low-fat diets, anxiety, isolation, and limitations in education and employment. Patient-reported studies document profound quality-of-life loss and disruption to work.[9,69]

Note: Acute pancreatitis admissions are resource-intensive across Europe, representing a substantial and avoidable burden on healthcare systems. Timely diagnosis is therefore critical to reduce both costs and preventable patient suffering.[9]

Diagnosis: Early genetic confirmation—crucial for distinguishing FCS from multifactorial hypertriglyceridaemia, shortens the diagnostic odyssey and enables tailored care pathways. Testing should be initiated whenever there is strong phenotypic suspicion.[9]

Prevention and management: Management requires a combination of a very-low-fat diet, regular specialist follow-up, and, where appropriate, innovative triglyceride-lowering therapies now available in Europe. Ensuring equitable access to both treatments and expert centres is key to reducing pancreatitis episodes, hospitalisations, and the broader societal costs.[10, 73,74,75]

Annex 2. Implementation

National Examples and Cross-Cutting Lessons from PERFECTO-FH29

The PERFECTO-FH meeting and existing data[70] showcase a wide range of national approaches to FH detection and screening across Europe, demonstrating that implementation is feasible in different healthcare systems when supported by political commitment, clinical leadership, registries, and collaboration.[48]

Examples of National Approaches

- **The Netherlands** demonstrated the long-term impact of nationwide cascade screening, showing major reductions in premature CVD across generations, while also highlighting the risks of stopping sustained national programmes.
- **Slovenia** presented one of the world's leading universal childhood FH screening programmes, integrated into mandatory preventive care at age 5, combined with genetic testing and reverse cascade screening of parents.
- **Czech Republic** showcased the long-running MEDPED programme alongside innovative pilot projects for newborn and toddler screening using genetic testing and structured referral pathways.
- **Denmark** demonstrated how national quality registries, mandatory reporting, performance indicators, and political engagement can drive nationwide implementation and accountability.

- **Croatia** combined preschool screening with large-scale public awareness campaigns, community outreach, and preventive cardiovascular health initiatives.
- **Luxembourg** implemented a national universal FH screening programme at 18 months of age using finger-prick testing and centralised referral systems.
- **Poland** presented integrated family-based care models combining paediatric and adult lipid clinics, genetic testing, registries, and structured transition pathways.
- **Latvia** showed how significant progress can be achieved even without dedicated national funding through clinician leadership, registries, GP education, digital tools, and EU project collaboration.
- **Portugal** shared important lessons from implementation challenges, highlighting the need for simple, scalable, GP-friendly screening pathways.
- **Cyprus** focused on integrating cardiovascular prevention and FH into broader national health reform and digital prevention strategies.
- **Ukraine** demonstrated how grassroots clinician-led initiatives and collaborative registry building can begin even under extremely challenging conditions.

Cross-Cutting Lessons

Across all country examples, several consistent implementation lessons emerged:

- **Early detection in childhood is critical** to prevent lifelong LDL-C exposure and premature ASCVD.
- **Cascade screening remains one of the most effective and cost-efficient strategies** for identifying affected relatives.
- **Primary care and paediatric engagement are essential** for scalable implementation.
- **National registries and digital infrastructure are key** for monitoring, quality improvement, and sustainability.
- **Genetic testing strengthens diagnosis and family tracing**, particularly in children and borderline cases.
- **Patient organisations play a crucial role** in awareness, advocacy, education, and policy engagement.
- **Public awareness campaigns improve participation and political visibility.**
- **Pilot projects are important starting points**, but long-term sustainability requires integration into national health systems and public funding.
- **Cross-sector collaboration between clinicians, policymakers, researchers, and patient organisations is essential** for successful implementation.

The PERFECTO-FH discussions demonstrated that Europe now has multiple real-world models proving that systematic FH detection and early prevention are achievable, scalable, and increasingly ready for wider implementation across EU Member States.

Medical Capacity Building: The EAS Lipid Clinic Network

Effective implementation of cardiovascular health checks requires not only policy recommendations but also clinical infrastructure capable of supporting awareness, education, early detection, referral pathways, family-based prevention, and long-term management. The European Atherosclerosis Society (EAS) Lipid Clinic Network represents an important implementation infrastructure already being developed to support these objectives across Europe and beyond.[76]

The Network currently connects more than 500 lipid clinics across over 60 countries and aims to strengthen harmonised approaches to screening, diagnosis, treatment, and long-term management of inherited lipid disorders, including FH and elevated Lp(a). It supports the development of specialised expertise, family cascade and reverse-cascade screening pathways, registries, and dissemination of best clinical practice across healthcare systems.

Importantly, the Network places strong emphasis on awareness raising, education and capacity building of clinicians and healthcare professionals. Through collaboration with national experts, multilingual educational activities, webinars and dissemination initiatives, the Network supports implementation of harmonised approaches adapted to different healthcare systems and local realities. This contributes to strengthening clinical expertise in primary care, paediatrics, cardiology, endocrinology and specialised lipid services.

The Network also recognises early paediatric detection as a key prevention priority. Children with inherited lipid disorders represent some of the highest lifetime cardiovascular risk groups, despite often remaining asymptomatic for decades. The Network therefore supports earlier identification of affected children and families through structured paediatric screening and family-based prevention approaches integrated into broader healthcare systems.

Together, these developments demonstrate that Europe is already building awareness, education, clinical expertise, and implementation infrastructure needed to support coordinated cardiovascular health checks, including early detection and lifelong management of inherited cardiovascular risk across Member States.

Digital Health Tools and the European Health Data Space (EHDS) for comprehensive health checks

Digital health technologies, artificial intelligence (AI), interoperable electronic health records (EHRs), and the European Health Data Space (EHDS) represent an important opportunity to strengthen early detection and prevention of inherited lipid disorders across Europe. EHR-based screening algorithms can support the identification of previously undiagnosed familial hypercholesterolaemia (FH) by systematically analysing routinely collected clinical data, including lipid profiles, medication history, diagnostic codes, and family history of premature cardiovascular disease.

Automated phenotyping and machine-learning tools, including the **FIND FH** algorithm and **Mayo SEARCH**, have demonstrated the ability to identify individuals at high risk of FH within large healthcare datasets, substantially improving detection of this underdiagnosed inherited condition. When integrated into clinical decision-support systems, EHR-based alerts can support earlier referral, genetic testing, treatment optimisation, and family-based cascade screening.[77, 78]

Evidence suggests that implementation should follow a strategic and risk-based approach. Targeted EHR screening in high-risk populations, such as cardiology or lipid clinics and patients with severe hypercholesterolaemia or premature cardiovascular disease, demonstrates very high positive predictive value while minimising unnecessary follow-up and clinician burden. By contrast, fully automated population-wide screening may generate large numbers of alerts with lower predictive value, increasing demands on healthcare systems and contributing to alert fatigue.

The implementation of EHDS creates a unique opportunity to improve interoperability, registries, cross-border data exchange, and secondary use of health data for cardiovascular prevention and public health planning. At the same time, important challenges remain, including fragmented health systems, incomplete family history data, limited structured clinical information, and the need for safeguards related to data protection, transparency, explainability, ethical AI governance, and equitable access.

Annex 3. Social determinants of health

Using the Dahlgren-Whitehead model, a number of disparities in FH screening in FH were explored. across different layers of society. In PERFECTO FH the framework was applied to real-world experiences of marginalised and vulnerable groups, particularly Roma in Romania and migrants and refugees in Cyprus, helping to translate abstract inequalities into practical entry points for action.[59-69]

- **Key disparities in FH** Research show that differences in FH diagnosis, treatment, and outcomes are **not only medical**. They are strongly shaped by **social, economic, and systemic factors**. Across these layers, several key disparities emerge:
- **Unequal access to FH diagnosis and treatment.** People who are **white, male, higher-income, and more educated** are more likely to be diagnosed and receive treatment.
- **Underrepresentation in research and data.** Clinical trials and FH registries often **underrepresent minority and low-income populations**, limiting accurate diagnosis and tailored care.
- **Barriers to genetic testing and screening.** Access is reduced by **costs, location, language barriers, and mistrust** in how genetic data is used, lowering **participation in screening and cascade testing**.
- **Low health literacy.** Around **1 in 5 people with FH** struggle to understand health information, with those from **lower-income** and **education categories** reporting the lowest levels.
- **Lower participation in cascade screening.** Families from disadvantaged backgrounds are **less likely to inform relatives or engage in follow-up testing**, limiting early detection.
- **Geographical and income-based inequalities.** People in **lower-income countries** are less likely to receive genetic diagnosis or treatment and have **higher cardiovascular risk and complications**.
- **Gender inequalities in care.** Women are often **diagnosed later**, treated less aggressively, and are more likely to experience **treatment gaps linked to reproductive health**.
- **Higher burden of risk factors in underserved groups.** Some populations face higher rates of **smoking, diabetes, and hypertension**, compounding FH-related risks.
- **Mistrust and limited outreach.** Negative past experiences and a lack of culturally appropriate communication lead to **lower trust in healthcare systems** and reduced engagement.

Community-based health mediation offers a practical and scalable approach to improving equitable access to FH screening and care among underserved populations. Through trained mediators working within communities, it helps strengthen trust, improve health literacy, support navigation of healthcare systems, and increase participation in screening, diagnosis, and long-term care. While not a substitute for structural reform, mediation provides an immediate mechanism to reach populations often underserved by existing prevention programmes, including Roma and migrant communities.

Building on experiences from across Europe, including Romania and Cyprus, PERFECTO FH identified key building blocks for more equitable FH screening:

- community-rooted mediators to strengthen trust and engagement;
- culturally adapted FH education and health literacy;
- support for communication between families and healthcare professionals;
- practical navigation of administrative, financial, and linguistic barriers;
- active outreach for paediatric and cascade screening;
- equity monitoring and community-level data collection; and
- integration with primary care, public health, and social support services.

Together, these approaches support earlier diagnosis, stronger community engagement, and more equitable cardiovascular prevention pathways across diverse populations. By building knowledge and trust, mediation strengthens communities' ability to engage independently with screening and care over time.

Combining these with the other PERFECTO FH tools, incl. the Personalised Communication Model (PCM) for tailored engagement, and the societal return on investment (sROI) framework for capturing wider social impact, equitable FH paediatric screening can be developed and delivered.

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