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Listening across health systems

Shared patient priorities for improving familial hypercholesterolaemia (FH) care in Europe

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Background

Familial hypercholesterolaemia is the most common inherited metabolic disease, affecting ~2.5 million people in Europe. It is associated with a high risk of premature heart disease, yet most cases remain undetected. Closing the gap between prevalence and detection requires solutions that the population would be willing to adopt. The EU-funded project, FH-EARLY, is developing three solutions for early diagnosis and management of FH: a genetic chip array for detecting the disease, a biomarker assay for risk assessment and an explainable AI solution for health profiling. All three are being co-created with patients.

Objectives

To embed the involvement of patients and families across the FH-EARLY project through a structured *Involvement protocol for FH patients and families* and to understand how people living with FH and their families experience and manage the condition, by:

1. Assessing FH-related priorities, lifestyle and engagement of patients and carers with health services;
2. Identifying barriers and enablers in relation to managing FH at home, at work and in the community;
3. Translating priorities into end-user requirements that guide the co-creation of the FH-EARLY innovations.

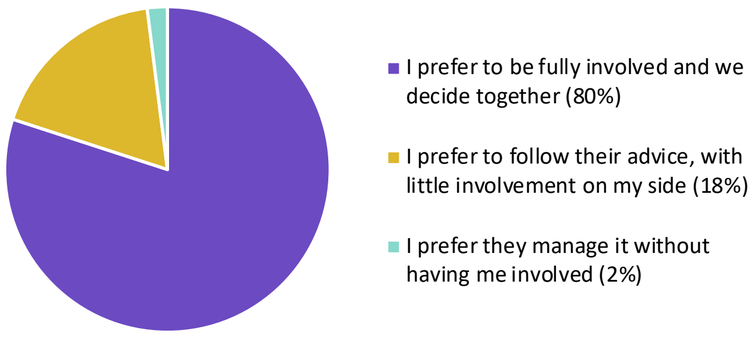
Methodology

A mixed-methods assessment guided by the *Involvement protocol for FH patients and families*, conducted between April-June 2026, implemented in two steps:

- Questionnaire on health-related knowledge and behaviour, information sources, support and attitudes towards novel technologies. 74 patients involved (Czechia=21, Portugal=45, France=8);
- Focus groups with 13 patients (Czechia=7, Portugal=3, France=3), using a common discussion guide covering FH diagnosis, co-management with doctors and family members, information needs, digital literacy, reaction to the three FH-EARLY solutions.

Results

Across all three sites, patients expressed their desire to manage FH together with their doctors, not around them. They want to be fully involved in managing FH and to share decisions with their healthcare professionals, not delegate them. Moreover, healthcare professionals are the first and the most trusted source of information for patients. Innovations should therefore reach patients through, and with the endorsement of, healthcare professionals.



Graph 1: *Preferred role in managing FH with healthcare professionals*



Graph 2: *Main sources of information when seeking advice about FH*

To some extent, **different priorities emerged across settings**:

- **Czech patients** described social dismissal of FH (including among HCPs), scarce reliable information and difficulty reaching specialist care: "*When I mention among friends that I have high cholesterol, everyone brushes it off - 'why are you telling us this, everyone has it'*" (female, 50); "*I kept going there asking to be referred for some examination. For several years she approached it dismissively - 'I've seen worse values'*" (female, 50, Czechia).
- **Portuguese patients** emphasised delayed recognition of FH, unclear care pathways and the absence of guidance on paediatric FH: "*I was followed in cardiology for many years and nobody ever requested a genetic test or asked about my family. It was assumed: she has 270 cholesterol, she's young, doesn't smoke, isn't obese, we'll just monitor*" (female, 47, Portugal).
- **French patients** highlighted treatment burden, side effects and uneven information: "*Whether to stop the statins to keep doing physical activity, or take them and accept reducing activity…in terms of patient education, it really was a dilemma*" (male, 66, France).

Despite these differences, **several priorities were shared across all settings**:

1) improving FH awareness, including among healthcare professionals: "*Frankly, I'm not even sure all cardiologists are familiar with this, because I was followed in cardiology for many years and nobody ever requested a genetic test or asked about my family*" (female, 47, Portugal);
2) promoting earlier diagnosis: "*We shouldn't have to wait until 40 to be diagnosed; it should be part of testing*" (male, 66, France);
3) strengthening cascade testing and structured follow-up for relatives;
4) receiving clear and credible information, that is consistent across the specialists involved in care;
5) recognising and actively managing treatment side effects: "*There's that thing where you start statins, your muscles hurt, you feel tired. But doctors usually just look at the LDL and say 'the LDL is great'. That sometimes weighs on us a bit as patients*" (male, 43, Portugal).

Conclusion

The gaps that patients describe concern not only access to diagnosis and treatment, but also navigation, communication and continuity of care. Earlier recognition of FH, screening, better informed healthcare professionals, clearer care pathways and systematic attention to treatment side effects should complement the advances in diagnosis and treatment. Two principles follow. First, patients do not want to be managed, they want to be partners. Across all sites they ask to be fully involved and to decide together with their clinicians, whom they trust above any other source. Second, the same holds for innovation: patients welcomed the FH-EARLY solutions, provided they are validated and introduced by trusted clinicians. If care and innovation are to reach people with FH, both must be built with them. Incorporating patient-identified priorities into care and into the development of new solutions is the route to more accessible, person-centred care.

Contact

For more details about the FH-EARLY project:

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