D5.1 Guidelines/models for citizens', patients', health professionals' engagement in Personalised Prevention: Report on the guidelines/models for citizens', patients', health professionals' engagement and communication to family members

Consisting of three sub-reports:

D5.1.A Guidelines/models for citizens', patients' engagement in Personalised Prevention

D5.1.B Guidelines/models for health professionals' engagement in Personalised Prevention

D5.1.C Communication to family members

VUMC/Amsterdam UMC. KUL. EPF & ACN

Funded by the European Union

PROPHET
a PeRsOnalized Prevention roadmap for the future HEalThcare

D.5.1.A Guidelines/models for citizens' and patients' engagement in Personalised Prevention

Engaging Public Voices: Towards Best Practices for Citizen and Patient Engagement in Personalised Prevention – qualitative study

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Deliverable Abstract

Introduction: Engaging public voices is essential for best practices in personalised prevention. We conducted a qualitative study to explore citizens' and patients' perspectives on engagement in personalised prevention, focusing on empowerment through education and communication.
Methods: In co-creation with the European Patients Forum (EPF) and Active Citizens Network (ACN) semi-structured interviews were conducted with 29 participants, including 11 patient representatives and 18 citizen representatives across 16 countries within Europe, representing (at least) seven distinct disease groups. Recruitment was conducted by EPF and ACN via newsletters and emails to their mailing lists. Thematic analysis identified key themes related to engagement and empowerment in personalised prevention across three domains: research, care and governance.
Results: Effective communication, via culturally sensitive approaches and diverse channels, was deemed crucial. Accessible information and educational resources, such as straightforward materials and online tools, tailored to the needs of patients and citizens, was considered important to support informed decisions in care and prevention. Enhancing digital literacy and health literacy among patients and healthcare providers was considered vital for better communication and empowerment. Participants highlighted the importance of holistic care, viewing citizens and patients as individuals with unique needs.
Discussion: Tailored communication and educational strategies are essential for effective personalised prevention. The study underscores the relevance of a holistic approach in personalised prevention, integrating mental health and overall well-being. This vision suggests healthcare providers, and policymakers to promote specialised training and diverse

communication methods to build trust and enhance citizen and patient engagement.

Keywords

patient engagement, public engagement, empowerment, personalised prevention, personalised medicine

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ACN	Active Citizens Network
EPF	European Patients Forum
PROPHET	Personalised prevention roadmap for the future healthcare
PM	Personalised Medicine
PP	Personalised Prevention

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1 Background

The vision of personalised prevention as part of a broader program of personalised medicine relies on having empowered and engaged patients and citizens that will be able to apply knowledge about their health and disease risk to prevent health problems from occurring or deteriorating.

According to the EU Joint Action on Patient Safety and Quality of Care (PaSQ), empowerment is defined as a multi-faceted process that helps individuals gain control over their own lives and expand their ability to take action on matters they deem significant (1, 2). In the realm of personalised prevention and healthcare, patient empowerment plays a pivotal role in enhancing care outcomes. According to Steele et al., patient empowerment is seen as a process of "activating" patients who, after rejecting the passivity of the sick role behaviour and taking ownership of their care, are "more aware of, satisfied with, and committed to their treatment regimens" (3).

If the public, patients, and families are to be active partners, then they must be systematically and meaningfully engaged in the planning, delivery and evaluation of the following domains relevant for personalised prevention: research, care and governance. In the research domain, the term public engagement has been used to characterise patient and public contributions to research via roles that range from "passive" study participants to "active" members of the public involved in all phases of the research (4), including agenda setting, prioritisation, selection of outcome measures, dissemination of results. In the care domain, the aim of public and patient engagement is to increase the public and patients' active participation in healthcare and prevention (5);(6). This may include for instance development of patient information and decision making on centers of expertise. Lastly, in the governance domain, the involvement of the public in decision-making processes regarding personalised prevention policies and programs, encompassing their participation in policy development, guideline formulation, and organisational governance structures (7). By actively engaging the public, governance processes become more transparent, accountable, and responsive to the needs and perspectives of the communities they serve (8).

It is imperative that the public comprehends and endorses this shift when personalised medicine and tailored prevention are incorporated into public health and the healthcare system. This study aims to carry out an exploration on how to best engage and empower citizens and patients in the field of personalised prevention, with regards to education and communication. Personalised prevention has been defined as aiming to 'prevent the onset, progression and recurrence of disease through the adoption of targeted interventions that consider the biological information (e.g. genetics, demographics, health condition), environmental and behavioural characteristics, the socio-economic and cultural context of individuals Citizen and patient views were explored through semi-structured interviews and focus groups to gather insights and elements for developing a best practice model for engagement in personalised prevention. This knowledge will be useful and informative in informing engagement activities in relation to personalised prevention. The development of elements of a best model for citizen and patient engagement is relevant for raising awareness among citizens and patients on the potential of personalised prevention in improving quality of life and health outcomes (9). It is critical to get first-hand feedback from citizens and patients regarding their experiences with and views on engagement and implementation of personalised prevention in Europe. Patient and citizen stakeholder engagement is essential to build a network of trust among physicians, researchers, clinical geneticists, patients, and citizens (5). This research highlights the crucial role of

providing understanding of how the public and patients can be empowered across the aforementioned domains of research, care and governance and aims to contribute to the successful implementation of personalised prevention program at local, regional and national levels.

Citizen and patient engagement is integral to the European project 'A PeRsOnalised Prevention roadmap for the future HEalThcare' (PROPHET) (PROPHET, n.d.). This interview study contributes to the PROPHET project's objective to co-create a Personalised Prevention Roadmap for the future healthcare with stakeholders, in order to support the definition and implementation of innovative, sustainable and high-quality personalised approaches that are effective in preventing chronic diseases (9).

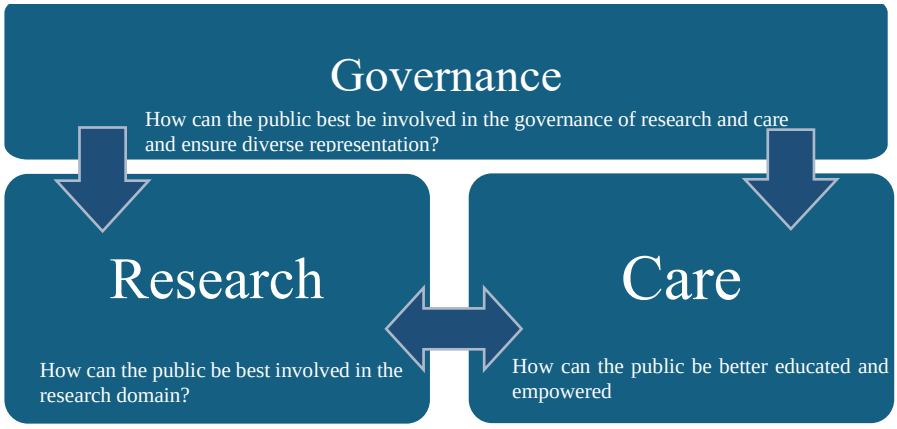
2 Methods

2.1 Study Design

A qualitative research strategy was chosen to obtain not only the opinion of the public on active and meaningful engagement in personalised prevention, but also to identify the rationale and explore views and experiences of the individuals informing their opinion. We ensured a mix of online focus groups (group-based approach) as well as individual interviews (one-to-one interviews) to explore best examples for engagement and empowerment based on effective communication and education according to the citizen and patient representatives across the three domains: research, care and governance.

The interview questions were formulated by the research team at Amsterdam UMC using the elements of PROPHET Deliverable 2.6: *Report on current practices of citizens' and patients' engagement in Personalised Prevention and their gaps/bottlenecks.* Key findings and topics from the Deliverable were extracted and discussed among the patients and citizens to understand which elements are useful and/or need further scrutiny in various practices related to prevention and personalised medicine. The discussions were conducted using a semi-structured interview protocol which included a range of questions across the three domains: research, care and governance (Figure 1). For each domain, participants were asked about best ways/strategies to improve engagement, communication, education and empowerment in personalised prevention; the key barriers and facilitators of public engagement in Personalised Prevention and Personalised Medicine, perception on the drivers of good engagement as well as preferences of engagement tools used and how to ensure diverse representation when engaging with the public (see Additional file 1 for the interview questions).

Figure 2: Domain Interactions within personalised prevention



The Medical Ethics Review Committee of Amsterdam University Medical Center evaluated the study design and decided that the Medical Research Involving Medical Subjects Act (WMO) does not apply to this study and that further official approval was not required (2024.014).

2.2 Participants

A purposive sampling strategy was used to recruit the participants. Recruitment was conducted by European Patients' Forum (EPF) and Active Citizens Network (ACN) via newsletters and emails to their mailing lists. At EPF, participants were assigned to either a focus group or an interview based on their role as patients or patient representatives. The goal was to organise two focus groups: one with patients and the other with patient representatives, ensuring that discussions were tailored to the specific perspectives and experiences of each group. At ACN, two groups of citizen representatives were formed. Prior to their scheduled session, participants received an informed consent form and a handout created by the research team (LLK, KG and CvE), providing background information and definitions of key terms. This preparatory material aimed to ensure participants were well-informed and could engage meaningfully in the discussions. The recruitment and scheduling process was handled by the EPF or ACN to ensure a high level of confidentiality, giving the researchers (LLK, KG and CvE) as little access to participant information as possible prior to the interviews or focus groups. Most participants from the qualitative study were older and highly educated. While patient representatives spoke on behalf of themselves and other patients, this group in particular is very active in health policy and advocacy work which means they likely have a deeper understanding of the health system issues and understand personalised medicine more than the average patient or citizen. The design, conduct and reporting of the study follows the Standards for Reporting Qualitative Research (10).The inclusion criteria for participation were an expressed interest in and prior experience with healthcare and preventive measures, ensuring we could build upon their knowledge while exploring new elements relevant for personalised prevention. In this study, the additional focus of personalised prevention was genetic information, though other avenues of personalisation such as via big data, were also discussed to a lesser extent. This approach allowed us to analyse the impact of genetic information on personalised prevention within the broader context of general healthcare, leveraging participants' existing familiarity with prevention and care. On completion, 29 participants took part in the discussions. We aimed to attain a range of perspectives across regions in Europe as well as the UK.

2.3 Data collection

The online interviews and focus groups took place on the Microsoft Teams platform. Six individual interviews were conducted, lasting between 45-60 minutes. Three online focus groups and one group discussion were conducted. Two focus groups were held with citizen representatives and two were conducted with patients and patients' representatives. The first citizen representative group *(n=8)* consisted of researchers, lawyer, administrator, pharmacist, physician, health manager, communication manager and a surgeon. The second group attended by citizen representatives *(n=7)* consisted of researchers, social scientists, nurse(s), nursing advocacy representative and a director of a patient organisation. Regarding the online patient (representatives) focus groups, one patient focus group was with patient representatives *(n=5)* and the other focus group/group discussion was with patients (*n=2*). Patients who could not attend the meeting for various reasons scheduled an individual interview. The patients *(n=11)* included came from a variety of (hereditary and non-hereditary) disease areas allowing the research to be broadly applicable rather than specific to one disease area. The focus groups lasted between 90-120 minutes. There were three categories of participants from the research

team in the focus groups: moderator, observer/technical-support, note-taker, while a representative from EPF or ACN who recruited the participants was present to welcome the participants.

Prior to the focus groups an interview was scheduled to check drafted questions. After the first focus group the introduction was simplified and the order of the domains was streamlined. For the individual interviews no slides were used but the introduction of personalised prevention and personalised medicine (See Appendix 1) and the questions per domain (See Appendix 2) were done verbally.

The data was stored locally on the server of Amsterdam UMC. The focus groups and individual interviews were conducted between 24th April 2024 and 05th of July 2024.

2.3.1 Feedback session/Member check

A feedback session (5 July 2024) was organised in collaboration with EPF and ACN to gather input on the preliminary results of the study. A total of eight participants took part in the discussion, comprising seven previously interviewed participants and one new participant from the PROPHET stakeholder forum. The participants included citizen representatives (n=4) and patient (representatives) (n=3), encompassing a diverse group with backgrounds in a Ministry of Health, citizen rights advocacy, research, patient rights association, patient safety, medical practice, and a migrant organisation. These participants represented interests and perspectives from local, national, and European levels.

The discussion aimed to validate the preliminary findings and explore several key areas, including identifying any missed areas in the study, distinguishing between the needs of citizens and patients, suggesting additional tools and instruments to enhance engagement and empowerment across diverse target groups, and clarifying the roles and responsibilities of professional organisations, governments, and other stakeholders.

The participants largely agreed with the preliminary findings, though they emphasised certain points for further consideration. This feedback was instrumental in refining the study's conclusions and recommendations.

2.4 Data Analysis

All discussions were held online, focus groups and interviews were audio recorded, and transcribed. Data were iteratively analysed using the MAX-QDA software version Plus 2022 (Release 22.1.1) to conduct thematic analysis. The coding analysis was performed across the individual interviews and focus groups. Initial coding, comprising open coding and category identification was performed by two researchers (LLK and KG) independently, in which the first two individual interviews guided the analysis of the rest of the interviews held. Thereafter, a final codebook was developed and discussed with the research group (LLK, KG and CvE). Based on the codebook, main themes and subthemes were derived and categorised in each domain.

3 Results

3.1 Participants

The characteristics of the 29 participants are shown in Table 1. The study encompassed participants from 16 countries of residence (see Figure 1), representing (at least) seven distinct disease groups. In total, the cohort included 11 patient representatives and 18 citizen representatives. Among the patients, the most prevalent conditions were Parkinson's disease

(*n=4)*, followed by various forms of cancer (*n=3)*, obesity (*n=1)* and diabetes (Type 1) (*n=1)*. Additionally, several patients mentioned that they had disorders in which genetics may have played a role, such as breast cancer (*n=2)*, rheumatic disease (*n=1)*, and hemophilia/Von Willebrand disease *(n=1)*. It is noteworthy that some citizen representatives, were also involved in health advocacy or personally affected by a disease, but chose to contribute from the perspective of a citizen (representative) or their professional experience. This distinction was explicitly communicated by these individuals.

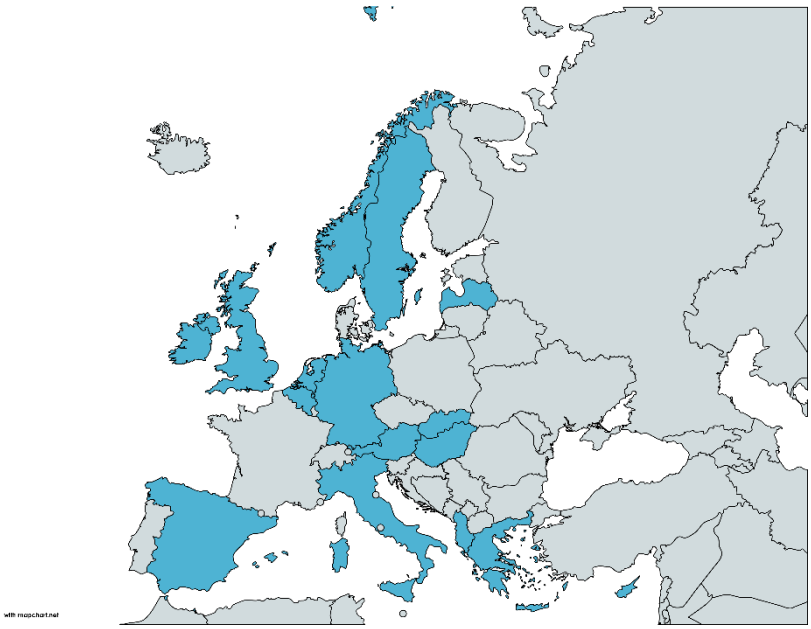


Figure 3: Representation across Europe

Table 1: Participant's country of residence

Country of residence[A-Z]	*n=29*
1. Albania	1
2. Austria	2
3. Belgium	3
4. Cyprus	1
5. Germany	1
6. Greece	2
7. Hungary	1
8. Ireland	2
9. Italy	4
10. Latvia	2
11. Netherlands	1
12. Norway	1
13. Slovakia	2
14. Spain	3
15. Sweden	1
16. UK	2

3.2 Themes

The qualitative findings in this study provide a comprehensive understanding of the various facets of citizen and patient engagement in personalised prevention. The analysis begins by exploring the motivations of the participants, highlighting their reasons for engaging in the research (4.2.1). It then examines the public's understanding of personalised prevention, assessing how participants perceive and interpret relevant concepts (4.2.2). Finally, the qualitative data is analysed across the three domains of research (4.2.3), care (4.3.4), and governance (4.2.5), highlighting key themes and insights. This approach offers a holistic view of the current landscape and identifies critical areas for improvement and best practices in engaging citizens and patients in personalised prevention. The last chapter provides an overview of the various engagement tools identified by the participants across the three domains.

3.2.1 Motivation to participate

Participants were primarily motivated to take part in the study by a desire to improve citizen and patient engagement in healthcare. Many participants were deeply involved in patient advocacy and healthcare organisations, driven by personal experiences with illness or professional roles in public health and patient safety. Their motivations included a keen interest in contributing to and learning about personalised prevention, advocating for patient rights, and improving healthcare access and outcomes.

> *"My interest in personalised prevention of chronic disease is because I think it has a deep impact on the affected person and the social realities, but also on the public health system… I'm [an] affected person myself."* (FG3citizenrepresentative)

> "*I was diagnosed with early onset Parkinson's….part of the treatment is looking at genetics…and there are quite a few different theories as to why it happens and how it progresses, so it really opens itself up to a personalised medicine approach. That peaked my interest." (Int1patient1)*

Others were interested in receiving information on personalised prevention based on experiences of their disease. Participants found the concept of personalised prevention both new and compelling. Some were particularly intrigued by the prevention component itself.

> *"Given the concerns regarding the sustainability of our economy and health service moving forward, we need to be preventing more ill health…My personal motivation to be on this call today is: how do we get top level ideas like prevention all the way down to individual behaviour change, and that's such a complex process… or maybe it's not."* (FG2 citizen representative).

These diverse perspectives leveraging personal experiences and professional expertise highlight the interest in personalised prevention, and expectations regarding its potential to transform individual health outcomes and broader public health systems.

3.2.2 Public Understanding of Personalised Prevention

Participants' knowledge of personalised medicine and personalised prevention came from healthcare providers, conferences, and various media. However, understanding varied, with some participants having detailed insights while others remained uncertain about practical applications. Many recognised the potential of personalised prevention to tailor treatments based on genetic information and individual risk factors, thereby improving health outcomes and resource allocation. This view illustrates a forward-thinking approach, seeing personalised prevention as an inevitable evolution in healthcare.

> *"I think personalised prevention is the inevitable future of health systems, especially because we now have new technologies that allow it, and also because we have a problem with resources. So it seems like the best solution overall... I'm curious to see how it can be translated in reality." (FG2 citizen representative).*

Participants discussed personalised prevention as a means to address resource limitations and enhance healthcare efficiency. A participant emphasised the importance of accurate testing to avoid false positives and negatives, which can impact patient trust and participation. Robust diagnostic tools in personalised prevention were seen as vital for effective and equitable healthcare.

> *"It's very important that people do not get false positive results that could impact their trust and willingness to participate in prevention programs in the future [...] it is also important that they do not get false negative results either, so that people who are at high risk are not left behind [because they don't have access to accurate tests]" (FG4 patient representative)*

Participants noted that better education and communication are needed to ensure the public fully understands its benefits and implications of personalised prevention. Despite some having firsthand experiences with genetic testing, gaps in knowledge about how personalised prevention could impact their overall healthcare were evident.

> *"I went through the process of doing genetic testing for my cancer; the doctor gave me much more information [about the test and personalised medicine but] not so much about prevention. How can personalised medicine help me depending on the results of the genetic testing and how*

might the checkups or the treatment plans change in the future based on the genetic results? (Int4 patient representative 4)

Participants also revealed concerns about the broader implications of personalised prevention on healthcare systems and social equity. Concerns about the shift from a solidarity-based healthcare system to one focused on individual risk were mentioned, with fears that personalised prevention could exacerbate healthcare inequalities.

Healthcare systems with healthcare insurance systems in Europe are traditionally based on solidarity. And I'm wondering...whether we are moving away from this solidarity principle and going rather to the individual risk calculations...I hope this will never happen in Europe, but you never know." (FG4 patient representative)

3.2.3 Research Domain

Participants in the study highlighted various aspects of communication and participation that need to be addressed to ensure that the public can fully grasp advances in personalised prevention.

3.2.3.1 Participation in Research (Design)

Patient and public engagement in medical research has progressed significantly in recent years. It refers to the inclusion of people with relevant patient experience in the design, conduct, and dissemination of research, to gather feedback and integrate e.g. outcomes that matter to patients, what would be of most benefit to them, understand what is acceptable to them, etc. Participants noted that many patients are motivated to participate in research due to their passion for advancing science or the lack of available cures for their conditions.

"I think that patients would be actually rather comfortable [participating in research] because, first of all, some of them are willing to help this research and the science on the one hand, and on the other hand, maybe their disease can't be cured. So they would be really interested in participating because they have actually no other choice [than the investigated drug]" (Int4 patient representative 21)

"There's very little training in medical school to teach us to speak science in common sense language... the better the scientist, the more able they are to convey what they have been learning in a layperson summary." (FG2 citizen representative)

3.2.3.2 Clarity and Accessibility of Information

One of the recurring themes was the necessity for clear and accessible information. Participants emphasised that complex medical jargon should be avoided to make the information more understandable to the general public.

There are a lot of talks about Google. I believe it is important that citizens are aware and informed, but not all information is correct on the Internet... I always advise people to talk because if there's a problem, you go on the Internet and you come out with a terrible impression, thinking that you're about to die. While it's probably a very simple issue. (Int6 citizen representative 28)

Participants also stressed the importance of respecting citizens' level of understanding in communication efforts. One participant highlighted the issue of "very technical information and communication" and the need for messages to be more understandable.

> *There are many super technical communications which the citizens cannot fully understand. It should be more targeted to the understanding of citizens. Citizens should be treated like adults. It is important to have better content rather than machine gun mass shooting of messages." (FG2 citizen representative)*

Effective public engagement within personalised prevention requires clear communication strategies that resonate with the general public's real-life experiences.

> *"So the lessons learned, how best to involve citizens? I believe that it works best when the language is understandable to the general public and close to their real-life experience."* (FG3 citizen representative)

> *"I am used to dealing with a lot of information from my background, but I found that it tends to be over the top... So sometimes, less is more" (Int1patient1)*

3.2.4 Care Domain

In the care domain activities aim to enhance quality of care and prevention by addressing individual needs and preferences through effective communication and education strategies, and supportive tools. Ideally, citizens and patients are well-informed, actively involved in their preventive care decisions, and supported throughout their healthcare journey. While patients often focus on care and secondary or tertiary prevention, citizens are primarily concerned with primary prevention, highlighting the distinct yet complementary needs within the domain.

3.2.4.1 Empowering Patients through Accessible Information and Effective Communication

Also for the care domain participants emphasised the importance of providing accessible and understandable information to empower individuals in personalised prevention. Effective communication is essential, yet there is a noted lack of training in medical schools on how to present complex information in layman's terms.

> *When I first knew that I had breast cancer I assumed it was one disease, not many kinds, and when I found out that I had the TNBC [triple-negative breast cancer; a rare type of breast cancer] there was no information on Norwegian websites[…]and the information I had was on big American sites...telling me that I was going to die very soon. So that was very scary. (Int5 patient 27)*

Participants also stressed the importance of tailoring communication strategies to the needs and preferences of the audience. One participant emphasised the role of social media and other communication channels in reaching and educating people, noting that

> *"I think that social media has taught us a lot about how to communicate or how to reach people. When doctors talk to patients, they are more prone to engage in the discussion. When we [patient advocates] communicate with citizens, they like short and understandable information." (FG2 citizen representative)*

Additionally, participants noted the challenge of balancing the right amount of information with the timing.

> *"It's a balance between the necessary and the right information at the right time... Having a lot of information when you're in a state of shock is no point [...] they don't tell you anything and expect you not to Google it, right?" (Int5 patient 27)*

Participants stressed the need for culturally sensitive communication and the use of diverse channels to effectively reach different demographic groups. They observed that younger individuals engage well with digital platforms like social media, while older adults tend to prefer face-to-face interactions. Improving digital literacy among both patients and healthcare providers is crucial for enhancing communication. Although younger patients often turn to online resources, older patients may require different strategies for effective engagement.

> *"We work with different ways of disseminating information through various channels. For young people, social media works more, but for older people, face-to-face interactions or advice in primary care centres work better. It's about using a combination of channels to communicate effectively with different groups." (Int6 citizen representative 28)*

The need to support establishing trustworthy, easily accessible sources of information that can help individuals make informed decisions about their health was highlighted by the respondents.

> *"...I think what we need on the national and even on the European level is a high-level platform with quality evidence-based information that patients, citizens and patient organisations can trust."* (FG2 citizen representative)

> *"My first step has been to make sure that I understand, so that I can make informed decisions when we're being asked to sign all these informed consent forms."* (FG2 citizen representative)

Providing reliable, evidence-based information and being transparent about the strengths, limitations and uncertainties of personalised prevention can help in gaining public trust.

These insights underscore the need for healthcare systems to adapt communication strategies to diverse needs and preferences, ensuring that all individuals have the information and support necessary to make informed health decisions.

3.2.4.2 Person in personalised prevention

Several participants indicated a need for a holistic care approach that transcends traditional disease treatment methods. This approach emphasises not only the physical health of individuals but also integrates their emotional, mental, and social well-being. Participants highlighted the significance of treating patients as a whole person and considering a range of factors that influence their health. The following quotes highlight gaps in current healthcare practices and suggest that a more holistic view could significantly enhance patient engagement and satisfaction.

> *"I really, really believe that patients are persons." (Int6 citizen representatives)*

> *"I think most of the points we made come down to more personalised medicine...seeing the person as a whole would really change the experience in the hospital...being able to read what*

you sign and the information linked to the treatment, that's the bare minimum of inclusion I think." (Int2 patient)

Participants in the study expressed the need for a holistic approach in personalised prevention, particularly highlighting the emotional and mental preparation required when receiving life-altering test results.

"When I realised how life-altering the results can be, I needed mental preparation and time... Despite initially agreeing to genetic testing for breast cancer, the complexity of the potential outcomes became evident during the waiting period, which coincided with my chemotherapy and exacerbated my mental health challenges. It's important for healthcare professionals to communicate on mental health...to take an extra step beyond the treatment plan." (Int2 patient)

A holistic approach also involves incorporating social determinants of health, such as environmental factors, cultural backgrounds, and community resources. Participants highlighted the need for a broader perspective that includes these elements to effectively engage and support patients. Addressing these factors is crucial for improving health outcomes and ensuring that prevention efforts are equitable and inclusive.

"We need to think in a more holistic approach including social environments, municipalities, culture, and other socio-determinants of health. We need to balance individual decisions, with individual data, but we also need to put in the predictive models based on environments and culture and other data that is not individual. (FM Int6 citizen representative)

3.2.4.3 Community Engagement and Support Network

Effectively reaching underserved or hard-to-reach populations requires engaging intermediary figures who have established trust within these communities. For instance, one participant highlighted the importance of involving those who already work closely with these groups underscoring the value of leveraging existing community connections to facilitate better outreach.

"*I think we need to go out and reach the people who never come to us. I think it's very important to count on the people who work with them as intermediaries...If I go to the prison... I have a lot of preliminary meetings with educators, with those who are reference points for these people." (Int6 citizen representative)*

It is also important to recognise that individuals are part of broader social networks that include family, friends, and community members, not just health professionals. Engaging intermediaries within these networks can help bridge gaps between health professionals and the wider community, ensuring that vital information about personalised prevention reaches diverse groups. The "mentor chat" system fosters community engagement by connecting mentors or peer supports who share experiences and offer psychological and informational support through online chats. This reduces patient isolation and promotes helpful exchanges See Appendix 3 for this tool and other specific strategies of successful community engagement practices

"It's important for patients to have a mentor—someone not necessarily a specialist, but willing to share experiences and support the patient. This helps patients realise they are not alone and can share lifehacks and information anytime, through a chat or community." (Int4 patient representative)

> *"We need to think that people are in the community with other people, with other relevant people and not only with health professionals... It's important to reach citizens, immigrants, and all people, focusing on primary prevention by working with municipalities and reaching out to those who are hard to reach." (FM Int6 citizen representative)*

Healthcare providers, especially general practitioners often serve as the initial point of contact for individuals and can significantly influence patient engagement. By focusing on smaller patient loads and specialised training, it was argued GPs can better educate and build trust with their patients, enhancing overall community engagement and support.

> *"I think empowerment and education of citizens involves general practitioners. They are at the frontline for citizens. You always go through your GP for anything.. and I believe they are increasingly well-trained now ... " (FG3 citizen representative)*

3.2.5 Governance Domain

Participants emphasised the importance of integrating public input into the governance and policy-making of personalised prevention. In the following, we will highlight roles and responsibilities for some key stakeholders, including governments, companies and healthcare professionals.

3.2.5.1. Challenges in Capability and Input

Participants mentioned significant challenges in terms of the capability and input within the governance domain, indicating a need for capacity-building and support for patient organisations to effectively participate in governance processes.

> *"[Governance is the] weakest of three domains in terms of input and capability." (Int1 patient 1)*

> *"How can this [stakeholder] involvement in governance be improved? I think through training, in schools for example or at the community level." (FG2 citizen representative)*

3.2.5.2. Structural Engagement and Regular Feedback

Participants stressed the necessity of regular, structured engagement opportunities where patient representatives can participate in discussions with policymakers and healthcare authorities. These sessions should include follow-up mechanisms to ensure that patient inputs are not only heard but also acted upon.

> *"And how to best engage patients? Invite the representatives of the patient organisations to the meeting, with the authorities, the Parliament, local committees or maybe with the Health Minister and to have round tables and discussions, and maybe to listen, not just to the matters which are on top [of the agenda]… what is important is to make these discussions regular, so with a follow up." (Int4 patient representative 21)*

3.2.5.3. (Personalised) Prevention higher on the agenda

Participants have regularly mentioned the necessity for governments to prioritise prevention in healthcare policies, moving from a reactive to a proactive approach. It underscores the potential of utilising data from various sources and fostering partnerships with citizens to enhance preventive measures. Additionally, it addresses the societal challenge of garnering interest in prevention before health issues arise.

> *"I think it's the obligation of the governments who organise the healthcare system to take care of patients' health status. The healthcare system is reactive and not proactive. There are a lot of data available in the healthcare system and also outside... We should channel this information into healthcare." (FM FG2citizen representative 10)*

> *"I think they [governments] are not focused on prevention. They focus on treatment itself because treatment is sexy and gets money. Prevention is not sexy and governments don't invest in prevention or work on prevention. So first of all, I think we need to change the government policies because the governments are shaping the programs." (FM_citizen representative 29)*

> *"If you are not a patient yet, you don't worry about your health, unless someone around you is sick. I think prevention is the hardest thing to achieve in society because of a lack of interest." (FM_citizen representative 29)*

These quotes underscore the need for governments to integrate prevention into healthcare policies by harnessing available data, engaging citizens actively in health management, and addressing the societal challenge of generating interest in preventive measures. An economic rationale was also mentioned for the government's obligation to ensure access to healthcare is critical.

> *"Treatment is more costly than prevention… From a health economics [standpoint], not allowing people to access to healthcare or not working on prevention costs healthcare systems more." (FM_citizen representative 29)*

In addition, healthcare professionals were thought to have a responsibility in promoting public health and prevention and the need for changes in medical education to support this.

> *"[prioritising prevention]… is not the responsibility of Coca Cola; it's the state's responsibility. First, it's the responsibility of the governments', then healthcare professionals. We also need to change the curriculum in medical school to better advocate for patient rights." (FM_citizen representative 29)*

3.2.5.4. Regulatory standards and data equity

In the feedback session, a participant highlighted the need for stringent standards regarding wearable technology and data collection practices. The importance of wearables being medically validated was emphasised, stating,

> *"When you think about wearables, we need to make sure they are medical grade, so you can actually coordinate the data that goes through." (FM FG2citizen representative7)*

> *"I agree there should be a quality assurance system for feeding information into health systems from various sources outside of health systems." (FM FG2 citizen representative 10)*

This assertion underscores the necessity for reliable and accurate data collection, particularly in the context of personalised health solutions. Additionally, a participant raised concerns about the lack of representation in data collection in EU member states, noting that

> "*We never* collect data *for non-white populations." (FM FG2 citizen representative 7)*

This gap can lead to adverse effects on these communities, highlighting the critical need for equitable data practices. In summary, all quotes collectively advocate for rigorous regulatory

standards and a commitment to data equity to ensure personalised prevention strategies are inclusive.

3.2.5.5. Collaboration with companies

The pivotal role of government in proactive health management has also been mentioned, the utilisation of available data for preventive measures, and the importance of partnerships between citizens and healthcare systems to enhance health outcomes. This emphasises the need for governments to take a more active role in disease prevention by leveraging data from various sources and establishing robust partnerships with citizens and other stakeholders such as private companies, ensuring that data protection rules adequately safeguard sensitive health data privacy without hindering healthcare advancements.

> *"I think healthcare data should be a means for the government to prevent diseases, to build prediction models. There should be a partnership with citizens because there are a lot of means through which patients generate data about their own healthcare like smartwatches... Health data protection is a huge issue, as data protection mechanisms prevent healthcare systems from exploring possibilities for improving citizens' health status." (FM FG2 citizen representative 10)*

> *"Private companies should allow citizens to transfer the information they collect to national health systems. This data could be a valuable source for the whole healthcare system, but it is not explored." (FM FG2 citizen representative 10)*

The role of private companies in the field of personalised prevention is crucial yet contentious. Participants highlighted the significant impact these companies have on the accessibility and affordability of preventive healthcare technologies. They emphasised the need for government intervention to regulate prices and ensure equitable access to essential medical devices.

> *"If we focus on the prevention part, there are millions of devices that focus on prevention which are quite helpful. But the price is not justifiable, it's very high. It's a monopoly of certain companies. It's the role of the government to make those devices massively available*[...] *private companies don't care about prevention, so the State responsibility first of all is working on prevention." (FM citizen representative 29)*

This underscores the need for regulatory frameworks and engagement with the private sector to enhance the accessibility of preventive healthcare technologies, ensuring that citizens can benefit from advancements in personalised prevention.

3.2.6 Diverse Representation and Outreach Strategies

Participants stressed the importance of inclusive governance and policy-making to address diverse populations in personalised prevention. Emphasising that effective governance must incorporate the experiences and needs of diverse groups to develop equitable strategies. Initiatives aimed at cultural sensitivity were particularly highlighted.

> *"Diversity and equity are central to effective personalised prevention. It's essential to consider cultural nuances and adapt healthcare practices accordingly," (Int4 patient representative 21)*

> *"We are starting to address inequalities in public health. We are focusing on vulnerable populations including people in prisons, immigrants, those living alone, and even illegal*

> *immigrants in our region. Our efforts include developing European projects aimed at economic ethics and chronic diseases, particularly focusing on methodologies that are cost-effective and sustainable within the healthcare system." (Int6 citizen representative 28)*

This sentiment underscores the necessity of engaging intermediaries such as educators and community leaders to bridge gaps and ensure inclusivity in healthcare initiatives.

Reflecting the importance of empowering patient communities and ensuring their voices are integrated into healthcare planning and decision-making.

> *"It should be more and more about Health democracy; patient communities really need to be seen and to be heard. And when we do it together, I really think we can see results. Everybody could be a patient at some point. So the quality of healthcare really affects all of us." (FG4 patient representative)*

Moreover, there was a recognition of the unique challenges faced by vulnerable populations. This highlights the imperative to tailor interventions that consider socio-economic factors and geographical disparities to ensure equitable access to personalised prevention strategies.

> *"In our region, for example, it's very important to engage all people, including the elderly and those living alone, who may lack access to transportation and internet services," (Int6 citizen representative 28)*

> *"Addressing the needs of diverse communities is crucial, especially considering differences in genetic predispositions, environmental influences, and lifestyle choices," (FG2 citizen representative)*

Participants addressed the responsibilities of both government and healthcare systems in reaching out to citizens, particularly through early intervention and culturally sensitive communication. It underscores the importance of proactive engagement with diverse communities to promote preventive health measures.

> *"The main thing is to start early and reach out to children and their parents about prevention and healthy lifestyle, especially with respect to migrants... communication is really the key." (FM FG4 citizen representative 26)*

3.2.7 Engagement tools

Across the three domains, research, care and governance, engaging patients and the public in personalised prevention requires effective communication and education tools and strategies. Participants identified several tools and approaches in enhancing patient and citizen engagement and empowerment. Appendix 3 displays the various tools that have worked best in the experience of the interviewees.

4 Discussion

The qualitative approach highlight several aspects essential for elements of best practice models of citizen and patient engagement in personalised prevention initiatives. Across all three domains of research, care and governance the theme of empowerment is central, which participants consistently emphasised as pivotal in their interactions with healthcare providers, researchers and policymakers. Empowerment builds on adequate information and

communication. For instance, participants expressed a strong desire for healthcare information that is not only accessible but also empowering, enabling them to make informed decisions about their health and prevention. This empowerment was underscored by the need for clear explanations of medical advancements and genetic testing implications, ensuring individuals can navigate complex health information with confidence. Furthermore, awareness of the diversity of patient and citizen stakeholders was noted as vital for ensuring that varied perspectives are included in research and practice and information is tailored to the needs of individuals and communities. It is crucial to highlight the cross-cutting themes of empowerment, clear communication, and awareness of diversity across the three domains. By integrating these themes, the effectiveness and inclusivity of personalised prevention strategies can be significantly enhanced. This approach ensures that strategies are responsive to the diverse needs of the citizens and patients.

Several themes emerged across the conducted focus groups and interviews, but some were more specific. In the care domain, the concept of patient-centered care was prominent, with participants appreciating healthcare practices that prioritise individual patient preferences, values, and needs. They valued providers who listened, understood their concerns, and involved them in decision-making, highlighting the need for a holistic approach that includes wellness and mental health. Challenges for healthcare personnel in communicating complex concepts and tailoring information to individual patients were noted, underscoring the need for adequate knowledge and skills to help patients take control of their health decisions. Beyond individual interactions, participants valued community-based approaches to personalised prevention, emphasising the role of support networks, patient advocacy groups, and community engagement initiatives - such as mentor chats, patient focused newsletters/ magazines and patient involvement and feedback in curriculum development (see appendix 3) - in disseminating health information and providing peer support. They stressed the importance of representatives from patient and citizens organisations maintaining strong community connections to ensure decisions reflect their needs. Additionally, the potential of digital devices and apps for ongoing monitoring and management of chronic conditions was highlighted, along with the importance of incorporating lifestyle assessments into patient care for more comprehensive and personalised health management.

Participants across all three domains emphasised the importance of integrating comprehensive health education into school curricula to teach basic health concepts and preventive care from an early age, promoting lifelong engagement in personalised prevention. Furthermore, establishing structured engagement and feedback mechanisms to ensure that citizens and patients can provide regular input on healthcare services and preventive measures, and involving community members in the planning and implementation of health initiatives to ensure their needs are addressed. Moreover, the integration of technology in healthcare delivery was noted also across all domains. Participants highlighted the potential of digital tools for improving access to health information, facilitating remote consultations, and enhancing patient monitoring in personalised prevention programs. Participants noted that while younger individuals are increasingly engaged in self-monitoring through health technologies and prefer digital communication, older individuals might have less interest or skills in digital tools, not technology savvy or experience and prefer traditional methods. This underscores the need for a balanced approach that accommodates both demographics to ensure inclusive access to personalised prevention resources.

4.1 Points to consider

Based on our findings, we suggest the following key points to consider across the three domains regarding the engagement of patients and the public in personalised prevention.

Table 2: Points to consider across the three domains regarding the engagement of patients and the public in personalised prevention

Research Domain - Points to consider

1. Raise awareness of the importance of and actively promote public involvement in research to improve research outcomes and develop personalised prevention solutions that address citizens' needs.
2. Develop accessible and empowering educational resources on medical advancements and genetic testing relevant for personalised prevention.
3. Integrate comprehensive health education e.g. via school curricula and provide ongoing community outreach activities.
4. Promote clear, jargon-free, communication from researchers and healthcare providers.
5. Encourage participation in research also for individuals and communities that may be difficult to reach, or hitherto not included in research.

Care Domain - Points to consider

1. Offer straightforward, jargon-free, health and genetic information to empower patients and support informed decision-making.
2. Collaborate with community intermediaries to effectively reach and support underserved or hard-to-reach populations.
3. Build trust through open, honest dialogue, about treatment options, risks, and personalised prevention strategies also through comprehensive and understandable informed consent forms.
4. Use digital platforms to increase access and engagement, ensuring that all patients benefit from technological advancements, while also making sure people who cannot a use such tools are also able to access care and information.
5. Tailor communication to meet the needs of diverse cultural, linguistic, and socio-economic groups.
6. Integrate holistic care approach that includes physical, emotional, mental and social well-being.
7. Adapt health messages to resonate with different demographic groups and individual preferences.

Governance Domain - Points to consider

1. Advocate for higher prioritisation of (personalised) prevention in policy agendas in relation to reactive healthcare models.
2. Establish structural engagement and regular feedback mechanisms to involve patients and community members in healthcare initiatives and policy-making.
3. Ensure diverse representation in healthcare initiatives to address the unique needs of different populations.
4. Integrate health prevention education into school curricula and provide ongoing community outreach activities, such as health workshops.
5. Implement communication strategies to reach diverse populations and build trust.
6. Provide and disseminate accessible, accurate, evidence-based information to the public in personalised prevention.
7. Support healthcare professionals' awareness of personalised prevention and ability to communicate with patients in an understandable way through specific training
8. Address the high cost of preventive devices e.g. through public-private partnerships and government interventions, and effective use of essential resources to promote equitable access to personalised prevention technologies.
9. Investigate the integration of data from wearable devices and building prediction models while ensuring robust privacy protections.

5 Strengths and Limitations

This study's strengths include the rich qualitative data obtained through in-depth interviews, providing nuanced insights into citizen and patient engagement in personalised prevention. The diverse participant pool, encompassing various stakeholders across multiple European countries, enhances the generalisability of the findings. The inclusion of participants from different disease areas further enriched the perspectives gathered. The active involvement of patient representatives, who are well versed in health policy and advocacy, added depth to the analysis. Practical recommendations for improving engagement, such as enhancing

communication, promoting health literacy, and strengthening community support, offer valuable guidance for policy and practice.

This study has several limitations. Sampling bias towards those already engaged in healthcare, potentially excluding less engaged or marginalized populations or citizens and patients, who are not members of a patient or citizen organisation, may have had an effect on results. While online discussions allowed broader geographical participation and comfort for patients, they excluded those lacking technological/digital literacy and non-verbal cues may have been missed, affecting interaction dynamics. Additionally, personalised prevention was sometimes interpreted as lifestyle changes rather than integrating wider data, such as genetic elements, possibly skewing findings. The demographic skew towards older, higher-educated participants and the use of English, a non-native language for most, may have limited the full expression of thoughts despite efforts to clarify misunderstandings.

6 Conclusion

Overall, the findings underscore the multifaceted nature of engaging public voices in personalised prevention. They demonstrate that empowering individuals through accessible information, transparent communication, and comprehensive health education can foster an environment where people feel confident and actively involved in managing their health. To advance personalised prevention effectively, it is crucial to develop policies that ensure systematic and meaningful engagement of patients and the public across the research, care, and governance domains.

A significant challenge identified is the general lack of knowledge and understanding of personalised preventive approaches among patients and citizens. Addressing this requires the provision of accurate, accessible information and educational resources, such as straightforward materials and online tools, tailored to the needs of patients and citizens.

Participants across all domains emphasised the importance of continuous, meaningful engagement throughout the entire healthcare process—from initial contact through to ongoing prevention, treatment, and evaluation. They stressed that clear communication about research processes, treatment options, and the implications of personalised prevention strategies—like genetic testing and lifestyle interventions—is vital for building trust and ensuring effective care and prevention. Such meaningful engagement and empowerment are integral to developing a best practice model in personalised prevention, fostering a more inclusive and responsive healthcare system where patient and citizen perspectives are actively integrated.

Healthcare providers and policymakers can create a supportive environment that empowers individuals to actively participate in their own health management while promoting equity and transparency in healthcare delivery of personalised prevention.

7 Funding

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9 References

1. PaSQ. European Union Network for Patient Safety and Quality of Care 2009 [Available from: www.duque.eu.
2. EPF. Campaign on Patient Empowerment: Roadmap for Action 2015 [Available from: https://view.officeapps.live.com/op/view.aspx?src=https%3A%2F%2Fwww.eu-patient.eu%2Fcontentassets%2F1f8150413eab4826be6f11b969bc0b63%2Fdraft-roadmap---patient-empowerment.docx&wdOrigin=BROWSELINK.
3. Steele DJ, Blackwell B, Gutmann MC, Jackson TC. Beyond advocacy: a review of the active patient concept. Patient Education and Counseling. 1987;10(1):3-23.
4. Schuster AL, Crossnohere NL, Paskett J, Thomas N, Hampel H, Ma Q, et al. Promoting patient engagement in cancer genomics research programs: an environmental scan. Frontiers in Genetics. 2023;14:1053613.
5. Pezzullo AM, Sassano M, Hoxhaj I, Pastorino R, Boccia S. Citizen engagement initiatives in precision health in the European Union member states: a scoping review. BMJ open. 2021;11(7):e045846.
6. Boote J, Telford R, Cooper C. Consumer involvement in health research: a review and research agenda. Health policy. 2002;61(2):213-36.
7. Onstwedder SM, Jansen ME, Leonardo Alves T, Cornel MC, Rigter T. Pursuing public health benefit within national genomic initiatives: learning from different policies. Frontiers in Genetics. 2022;13:865799.
8. Rijal S. The importance of community involvement in public management planning and decision-making processes. Journal of Contemporary Administration and Management (ADMAN). 2023;1(2):84-92.
9. PROPHET. a Personalised Prevention roadmap for the future HEalThcare n.d. [Available from: https://prophetproject.eu/.
10. O'Brien BC, Harris IB, Beckman TJ, Reed DA, Cook DA. Standards for reporting qualitative research: a synthesis of recommendations. Acad Med. 2014;89(9):1245-51.
11. De Paoli P, Ciliberto G, Ferrarini M, Pelicci P, Dellabona P, De Lorenzo F, et al. Alliance Against Cancer, the network of Italian cancer centers bridging research and care. J Transl Med. 2015;13:360.

10 Supplementary Material

Appendix 1: Participant Handout provided to respondents prior to the interviews and focus groups.

Participant Handout: PROPHET Personalised Prevention Discussion

Defining guidelines and best models for citizen and patient engagement

Thank you in advance for your time and participation in this study about your experiences and thoughts on citizen and patient engagement in personalised prevention. This handout will provide some background information on the topics we will discuss together.

Purpose of this discussion

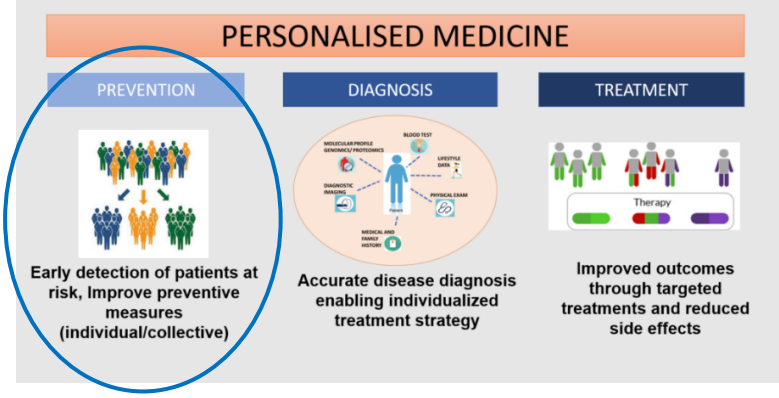
The purpose of this discussion is to help develop better guidelines for engaging citizens and patients in implementing personalised prevention throughout the European Union, to ensure that their needs and concerns are reflected.

We encourage you to think about and reflect on your personal experiences relating to personalised medicine and personalised prevention ahead of the meeting. Given your experiences, we hope to ask about how to communicate about personalised prevention, barriers to using personalised prevention, and ideas for improvements in citizen and patient engagement. We will also ask you about (ethical) concerns you may have.

List of terms

Here are a few concepts we will use during the discussion:

- **Genomics**: the study of all of a person's genes, including the interaction of those genes with each other and with the environment[i]
- **Personalised Medicine**: Personalised medicine (PM) is a form of medicine where a doctor uses information about the person's genetic profile and other data to guide decision-making about prevention, diagnosis and treatment. The idea is that you tailor "the right therapeutic strategy for the right person, at the right time." [ii],[iii],[iv]



- **Personalised Prevention:** Personalised prevention is a form of PM. "Personalised prevention aims to prevent onset, progression and recurrence of diseases through the

adoption of targeted interventions that consider the biological information*, environmental and behavioral characteristics, socio-economic and cultural context of individuals. This should be timely, effective and equitable in order to maintain the best possible balance in lifetime health trajectory"[v]

- * e.g. genetic and other biomarkers, demographics, health conditions

How do personalised medicine and personalised prevention connect? Every person has a unique health history. More concretely, personalised medicine and prevention come together to give tailored treatment and prevention recommendations based on a person's genetics *as well as* their overall health history and environment. For example, breast cancer can be caused by many different genes, environmental influences, demographics, and other previous health conditions[vi]. While breast cancer screening is generally offered based on age for the general population, families with higher genetic risks are seen in the clinic. Screening can be adapted (personalised), for instance start at an earlier age or be done more frequently, depending on the risk group.

- **Engagement**: There are many ways to define engagement, but for our discussion, we use engagement to mean:
  - At the Individual level: "the extent to which patients, [citizens] and their families or caregivers, whenever appropriate, participate in decisions related to their condition (e.g. through shared decision-making, self management) and contribute to organizational learning through their specific experience"
  - At the Collective level: "the extent to which patients [and citizens], through their representative organizations, contribute to shaping the health care system through involvement in health care policy-making, organization, design and delivery."[vii]
  - Why engagement? Engagement is important because it gives citizens and patients a voice in naming their concerns, priorities, and desires. The objective of engagement is to ensure that healthcare and health care policies meet the needs of those who use and receive these services, namely patients and the public. Engagement goes hand in hand with access to information, which is a key aspect of patients' and citizens' empowerment.

- **Empowerment**: Like engagement, empowerment has many meanings. In our discussion, we refer to empowerment as a: "process that helps people gain control over their own lives and increases their capacity to act on issues that they themselves define as important." [viii]

Explanation of Domains

During our discussion, we will explore three domains related to the implementation of personalised prevention, namely: research, care, and governance/policy-making. Below are some explanations with sub-questions about the different domains. In each of these domains, patients and citizens can be involved in personalised prevention practices in different ways. There are overlaps between these domains so we understand if you may see connections and/or similarities between these three domains.

Research: Involvement of patients and citizens in research decision making, e.g. setting research agenda priorities, and communicating research to the public

- How best to involve patients and citizens?
  - Examples of what works, what can be improved?

        - E.g., engagement tools, level of involvement (high/low), barriers to engagement
    - Improving communication about research

Care: Increase the public and patients' active participation in healthcare. This includes: communicating risks, such as genetic test results, improving health literacy.

- How best to educate and empower patients and citizens in healthcare?
    - Examples of what works, what can be improved?
        - E.g., educational tools, level of involvement (high/low), barriers to empowerment

Governance: Participatory decision making in health policy (having a seat at the table)

- How to establish trust?
    - Examples of what works, what can be improved?
        - E.g., engagement tools, level of involvement (high/low), barriers to engagement
    - How to make sure all voices are heard?

PROPHET Project

This research is part of the PROPHET project: a Personalised Prevention roadmap for the future HEalThcare. The PROPHET project is an European wide collaboration to create a roadmap, together with input from a broad range of stakeholders, including citizens, patients, healthcare professionals, and policy makers, to help implement personalised prevention.
More information about the PROPHET project can be found at this link: Home - PROPHET (prophetproject.eu)

[i] Source: https://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics https://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics
[ii] Source: https://www.genome.gov/genetics-glossary/Personalised-Medicine
[iii] Source: Personalised medicine - European Commission (europa.eu)
[iv] Image source: What is Personalised Medicine? (eulac-permed.eu)
[v] Source: Methodology section - PROPHET (prophetproject.eu)
[vi] Source: Breast Cancer Risk Factors You Can't Change | American Cancer Society
[vii] Source: https://www.eu-patient.eu/globalassets/campaign-patient-empowerment/epf_briefing_patientempowerment_2015.pdf
[viii] Source: https://www.eu-patient.eu/policy/Policy/patient-empowerment/

Appendix 2a: Interview questions for citizen representative

Welcome & Introduction
I would like to start with introducing myself and our team (mention names, institutions) and PROPHET briefly.
Then we will now start the recording.
Could you please mention:

- Name
- Where you are from
- Do you represent an organization?
- Motivation to participate

Brief explanation of PROPHET project:
Develop research and innovation agenda for personalised prevention, with many stakeholders, such as patient organisations, universities.
And our task is to study patient and citizen engagement, so we do this as the Amsterdam University medical center together with patient representatives EPF and ACN.

Personalised medicine aims to tailor health care and prevention more to individual needs and characteristics, away from the one size fits all approach. Many definitions exist, but the core is to also include genetic information, in addition to other types of data on life style, and also individual preferences or values.
Ideally **patients would play an active role** in their own health management, so would need to understand that genetic information is available and discuss health care options with their health care providers, e.g. via shared decision making, potentially contribute health data to further research.

Summary of Purpose:
As we described in the information package we're conducting this interview to hear your perspective on personalised prevention. We're particularly interested in your views on how patients and the public can be better engaged and empowered in this area.
Your input will contribute to developing best practices for patient engagement in personalised prevention.
House Rules

- Checking consent form, confidentiality - will treat the information confidentially, we will not use names in the reports.
- Have you had a chance to go through the handout?
- You will be asked a series of questions for discussion
    - Please feel you can speak openly and honestly
    - No right or wrong answers– we're simply interested in your experiences and opinions.

Do you have any questions at this point ?
Interview Overview:
During the interview, we'll be asking you a series of questions covering three main areas: research, care, and governance. They all relate to how best to engage citizens in personalised prevention/medicine
Questions per Domain
Care domain: How can citizens be better educated and empowered?

A. Your lessons learned:
How to empower and educate citizens to be prepared for personalised prevention
Can you describe one example that worked well, addressing e.g.:
1. What tool worked best, why?
2. How can this be improved?
3. What is missing?

B. Is there a difference between patient or citizen representation?

C. How can citizens best be involved in governance of care in personalised prevention?
1. What worked best, why?
2. How to ensure diverse representation?

Research domain: How are citizens involved?

A. Your lessons learned:
How best to involve citizens on setting e.g. research topics, priorities, design etc.
Can you describe one example that worked well, addressing e.g.:
1. What tool worked best, why?
2. How can this be improved?

B. How best to communicate about research to citizens:
1. What tools worked best, why?
2. Is information understandable / accessible?

C. How can citizens best be involved in governance of research in personalised prevention?
1. What worked best, why?
2. How to ensure diverse representation?

Wrap-up question
1. What are the two most important things that you think need attention in empowering citizens in personalised prevention/personalised medicine?

Thank you for your participation!
Closing & PROPHET follow-up:
- PROPHET Report will be made and distributed beginning of September 2024
  - Academic publication 2025-2026
  - VU-University/ AmsterdamUMC Master thesis on ethical aspects August 2024
- Input to PROPHET Capacity building for Public and Citizen engagement ACN & EPF 2025-2026
- Part of PROPHET Road map for Personalised Prevention in EU September 2026

Appendix 2b: Interview questions for the patient representatives

Welcome & Introduction
I would like to start with introducing myself and our team (mention names, institutions) and PROPHET briefly.
Then we will now start the recording.
Could you please mention:
- Name
- Where you are from
- Do you represent an organization?
- Motivation to participate

Brief explanation of PROPHET project:
Develop research and innovation agenda for personalised prevention, with many stakeholders, such as patient organisations, universities.
And our task is to study patient and citizen engagement, so we do this as the Amsterdam University medical center together with patient representatives EPF and ACN.

Personalised medicine aims to tailor health care and prevention more to individual needs and characteristics, away from the one size fits all approach. Many definitions exist, but the core is to also include genetic information, in addition to other types of data on life style, and also individual preferences or values.
Ideally patients would play an active role in their own health management, so would need to understand that genetic information is available and discuss health care options with their health care providers, e.g. via shared decision making, potentially contribute health data to further research.

Summary of Purpose:
As we described in the information package we're conducting this interview to hear your perspective on personalised prevention. We're particularly interested in your views on how patients and the public can be better engaged and empowered in this area.
Your input will contribute to developing best practices for patient engagement in personalised prevention.
House Rules

- Checking consent form, confidentiality - will treat the information confidentially, we will not use names in the reports.
- Have you had a chance to go through the handout?
- You will be asked a series of questions for discussion
    - Please feel you can speak openly and honestly
    - No right or wrong answers– we're simply interested in your experiences and opinions.

Do you have any questions at this point ?
Interview Overview:
During the interview, we'll be asking you a series of questions covering three main areas: research, care, and governance.
Questions per Domain
Care domain: How can patients be empowered and educated?

A. Your lessons learned from personalised prevention/personalised medicine:
   What is your view or experience with improving care & patient outcomes
   Can you describe one example that <u>worked well</u>, addressing e.g.:
    1. What tool worked best, why?
    2. How can this be improved?
    3. What is missing?

B. Do you have an example of effective communication tools? Or what could be done to improve communication by healthcare professionals (easy to understand content, toolkits, meetings, etc.)?
B. Is there a difference between patient or citizen representation?
C. How can patients best be involved in governance of care in personalised prevention?
    1. What worked best, why?
    2. How to ensure diverse representation?

Do you know of patients who were offered a genetic test as part of their care pathway, how did they feel about that?

Carefully probe:

-would you have want to know before,

-would it have changed your lifestyle, if prevention is possible

Research domain: how to improve patients involvement?

A. Your lessons learned personalised prevention/personalised medicine:
   How best to involve patients on setting e.g. research topics, priorities, design etc.
   Can you describe one example that worked well, addressing e.g.:
      1. What tool worked best, why?
      2. How can this be improved?

B. How best to communicate research findings/ research to patients:
   1. What tools worked best, why?
   2. Is information understandable / accessible?

C. How can patients best be involved in governance of research in personalised prevention?
   1. What worked best, why?
   2. How to ensure diverse representation?

If not addressed yet:
Do you feel comfortable participating in research (check: sharing your health data)
Do you feel comfortable participating in genetic research (check: sharing genetic data)

Wrap-up question

2. What are the two or three most important things that you think need attention?

Thank you for your participation!
Closing & PROPHET follow-up

- PROPHET Report will be ready September 2024
  - Academic publication 2025-2026
  - VU-University/ AmsterdamUMC Master thesis on ethical aspects August 2024
- Input to PROPHET Capacity building for Patient and Citizen engagement ACN & EPF 2025-2026
- Part of PROPHET Road map for Personalised Prevention in EU September 2026

Appendix 3: Engagement Tools across the Domains

Engagement tools	Domain	Purpose	Exemplar quotes	Explanation
Mentor Chat	Research and Care	Community enhancement/ Psychological and informational support	*"It's important for patients to have a mentor—someone not necessarily a specialist, but willing to share experiences and support the patient. This helps patients realise they are not alone and can share lifehacks and information anytime, through a chat or community."* (Int4patientrepresentative21)	The "mentor chat" system involves having a mentor or peer support system where patients can share experiences, receive support, and avoid feeling isolated through an online chat platform.

Patient Focused Newsletters/ Magazine	Care	Information and awareness	*"Newsletters mostly, and every member has a newsletter. Every local association connected to the big ones gets their own newsletters where we're meant to discuss it locally* (Int5patient27) *"I also put an article in […] magazine so everybody can read it. Individuals they were saying that it was really understandably written. I really ask the questions myself as I am also [...] patients. Yeah, I follow a lot of [...] research.[...] also on the genetic side and add it in my magazine"(FG1patient2)*	Patient-focused events and regular communication through newsletters are crucial for keeping patients informed and engaged. These events provide valuable information about ongoing research, treatments, and patient experiences, while newsletters help maintain a continuous flow of information, fostering a sense of community and engagement within patient groups.
Patient Involvement and Feedback in Curriculum Development	Care	Incorporating Feedback	*"There are opportunities to be involved in different ways through the university. I'm on the education teaching side and have been involved in the review of curriculum for upcoming medical students, helping to give patient feedback into the design... . It's important to get patient feedback rather than just raw research "* (FG1patient3)	Including patient feedback in the education of future healthcare professionals can improve the relevance and impact of medical training. Ensuring that patient perspectives are integrated into the curriculum of medical education can improve the quality of care and communication with patients.

ROPHET

a PeRsOnalized Prevention roadmap for the future HEalThcare

D5.1.B Guidelines/models for health professionals' engagement in Personalised Prevention

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PROPHET a PeRsOnalized Prevention roadmap for the future HEalThcare
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1	27-8-2024	Maria Luís Cardoso, Alexandra Costa, Astrid Vicente (INSA)	General feedback and input wording and clarification

Deliverable Abstract
For personalised prevention to become more integrated in health care systems, non-genetic ('mainstream') healthcare professionals need adequate knowledge to use and communicate about personalised strategies for prevention. Genetic education provides an important basis for such personalised strategies, as genetic information may be combined with other data on lifestyle, behaviour and environmental factors to optimise prevention. In this report we will discuss several challenges relevant for education when non-genetic healthcare professionals increasingly will be using genetic information as part of mainstream care and prevention pathways. It is important to address in genetic education to non-genetic healthcare professionals how standards from clinical genetics regarding e.g. counselling and potential risk for family members can be incorporated and further developed to be applicable in specific care and prevention domains relevant for primary, secondary and tertiary prevention. In discussions on guidelines points to consider include the need for continued genetic education for medical professionals, the crucial role of access to genetic expertise, clear protocols to establish roles and responsibilities, and the development of information and communication technology to support clinical decisions.

Keywords
Genetic education, non-genetic health care professional, mainstream care

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Table of Acronyms

CDS	Clinical Decision Support
CPIC	Clinical Pharmacogenetics Implementation Consortium
DPWG	Dutch Pharmacogenetics Working Group
FH	Familial Hypercholesterolaemia
GP	General Practitioner
ICT	**Information Communication Technology**
IHC test	**Immunohistochemistry test**
MRI	Magnetic Resonance Imaging
PM	Personalised Medicine

PP	Personalised Prevention
PROPHET	Personalised prevention roadmap for the future healthcare
VUS	Variants of Unknown Significance

1.Introduction

In a previous mapping exercise for the PROPHET project (Deliverable D.2.6 *Report on current practices of citizens', patients' health professionals and policymakers engagement in Personalised Prevention and their gaps/bottlenecks, part B: Mapping educational needs on personalized prevention for health care professionals: a narrative review)* we explored the educational needs of healthcare professionals regarding personalised prevention as a key component of engaging healthcare providers. That deliverable provided an overview of the knowledge and competencies required by non-genetic healthcare professionals to support the effective implementation of personalised prevention. It highlighted essential skills and competencies needed, such as utilising family medical history, given new developments in personalised medicine, and increasing 'mainstreaming' of genetics, allowing non-genetic healthcare providers to request genetic testing and/or perform counselling. We focused primarily on chronic conditions, such as hereditary cancers or cardiovascular disorders, which are also relevant to public health. Examples of education addressing relevant skills and knowledge across primary, secondary and tertiary prevention were discussed, along withmodes of education (face-to-face or online) and methods for evaluating such educational outcomes. The emphasis was on shifting from acquiring knowledge to being competent and trusted to apply the knowledge in practice.

In this new report (D.5.1.B), we aim to provide more background to general challenges and present key points to consider for developing guidelines regarding genetic education for non-genetic healthcare professionals, specifically focusing on increasing mainstreaming of genetic services relevant to personalised prevention. We do not aim to focus on evaluation of genetic education, for which recently a framework has been developed [1].

This report is based on literature and additional interviews with healthcare professionals actively teaching genetics to healthcare professionals.

Following an introduction to the mainstreaming of genetic care and prevention, we will first discuss some general overarching challenges relevant to educating non-genetic healthcare professionals in mainstream care and prevention. Subsequently, we will elaborate on three settings to illustrate developments and challenges for educating non-genetic healthcare

professionals in primary, secondary and tertiary prevention, including screening and pharmacogenomics.
Finally, we will conclude with points to consider for guidelines regarding genetic education for non-genetic health professionals, given the increasing mainstreaming of genetic services in care and prevention.

2. 'Mainstreaming' genetics for personalised prevention and challenges for education

In recent decades genetic information has increasingly become relevant for care pathways and prevention. Genetic education for non-genetic health care providers initially focused on recognition of hereditary disorders and knowledge when and how to refer to clinical genetics experts. In recent years a next step has been taken in allowing non-genetic health care providers, under certain conditions, to perform certain tasks of genetic services themselves. Treating physicians and healthcare providers, such as surgeons and oncologists, were allowed to, most notably, offer genetic testing themselves, and would refer patients to genetic services when test results would come back positive. For chronic diseases early implementation was in hereditary breast and ovarian cancer care. In this care domain a high caseload motivated care providers to an efficient use of resources. In addition, a significant part of the presenting cases was sufficiently similar to allow for a clear protocol, allowing these relatively straightforward cases to be tested for *BRCA* variants [2]. Similarly, for colorectal and endometrium cancer patients, pathologists increasingly started conducting routine or reflex immunohistochemistry (IHC) tests on tumours to screen for potential Lynch syndrome patients, that could be identified after subsequent germline testing [3]. Also in this case, further counselling was deferred to clinical geneticists. In these examples of autosomal dominant disorders, genetic testing could be used to adapt treatment and secondary prevention in an index patient, e.g. by more extended surgery or monitoring. In addition, first-degree family members could be counselled as they have a 50% chance of also carrying the same mutation, allowing for prevention and early interventions in a wider circle of family members at high risk of developing hereditary disorders.

This more straightforward pathway to mainstreaming genetics differs from other, more complex forms of mainstreaming found in highly specialised areas, such as neurology and cardiology. In these areas, specialists have increasingly recognised the genetic underpinnings of disorders in their patients, enabling them to order genetic testing and discuss potential diagnoses. For example, in cardiology, it may not always be possible to find a causal mutation, requiring counselling and monitoring of family members over time by the cardiologist.

Conversely, close collaborations between geneticists and cardiologists have developed, and subspecialties such as cardiogenomics have emerged.

It should be emphasized that in principle genetic expertise is certified and responsibility for genetic testing is in the hands of clinical or medical geneticists, while in some European countries also genetic counsellors and genetic nurses collaborate in patient care [4, 5]. Some countries provide more options than others for mainstreaming, and development of mainstream pathways is still in the early stages. If and under what circumstances parts of genetic services can be mainstreamed is still debated [6, 7]. Roles and responsibilities need to be clear to allow for efficient and responsible care, and shifting tasks require transparent communication and coordination. Against this background the two broadly painted scenarios for mainstreaming of genetic testing by non-genetic healthcare professionals come with various challenges for genetic education. In the following we will discuss some of the challenges for mainstream scenarios.

Pre-test counselling and obtaining consent for genetic testing

Informing and counselling patients before genetic testing is crucial to enable truly informed consent [8, 9]. This process can be challenging especially when the patient is already coping with a serious disorder and treatment decisions need to be made that can be informed by the result of the genetic test. Genetic education in mainstream care should address how healthcare professionals can inform and counsel patients adequately. Ideas on what is adequate, and attainable, in this situation, may differ between mainstream health care providers and clinical geneticists, who come from different practice backgrounds. For instance consultations cannot take up as much time as is available in clinical genetics. Bunnik et al. [9] argues in consenting for cancer care three types of unsolicited outcomes need to be discussed by mainstream care providers as they may have clinical and psychosocial consequences: suspected germline mutations, potential variants of unknown significance (VUS) and unsolicited findings pertaining to other conditions. It is important pre-test counselling in mainstream care should also address potential risk or consequences for first-degree family members in case of finding a pathogenic mutation, otherwise this message may come as an unexpected surprise when the patient receives the test results [8]. “The loss of the psychotherapeutic pre-test component when mainstreaming genomic testing risks increasing decisional regret and the related psychosocial burdens for the patient, as well as the chance of ethical and legal complications. Truly informed consent is a highly complex process which requires special consideration in genomic practice.”[8]
However, time constraints may not always allow for addressing potential familial consequences, and it could be argued that in many cases the test result would come back negative. Health professionals might argue that such familial consequences could be discussed during the disclosure of the test results.
In addition, although consent is needed for any medical procedure or intervention, treatment-related genetic testing may be perceived by treating physicians more as a standard test to ensure

adequate treatment, and less as part of a genetic care pathway. It will depend on the disorder and type of testing what tradeoffs between standards/ideals in clinical genetics regarding pre-test counselling and consent and mainstream care will be possible[10].

Consequences for family members

The request to perform a genetic test is part of a patient pathway that may have consequences for family members, as they might also be at risk and can take measures to prevent or delay the familial disorder. If the test results are communicated by the health professional it may not always be clear whether the potential risk for first-degree family members and the need to inform the family is discussed with the patient. Such aspects would be routine in a clinical genetics consultation. Therefore, if patients have a positive test result and are not referred to the genetics department, health professionals should be aware and knowledgeable on counselling the patient regarding the need to inform family members. It is crucial to make sure healthcare providers are in agreement on who is responsible for discussing consequences for family members, which is traditionally seen as a key task of clinical geneticists. Currently, the uptake of genetic testing by first-degree family members, after being informed, is approximately 40 % for hereditary cancers and cardiovascular disorders [11, 12]. Several factors contribute to this suboptimal uptake. For instance, personal or family-related aspects may also play a role: patients might not fully grasp the relevance of genetic testing or be willing, or able to inform their family members about contacting clinical genetics services. Additionally, family members may be hesitant, and accessing services can be challenging due to to distance or financial constraints. The third report of this Deliverable (D5.1.C) further explores the challenges for communication with family members.

Germline testing

When screening tests are being performed in mainstream care, such as in the case of Lynch syndrome tumours, follow-up genetic testing for germline mutations is not guaranteed, if the healthcare provider does not take action to refer to the clinical geneticist, or requests such testing himself, when allowed. For instance, in a recent review of reflex testing via IHC tests in mainstream care for Lynch syndrome in endometrial and ovarian cancer cases in the USA, uptake of germline testing was reported to be low at less than 40% [13]. Barriers may be identified at various levels. ‘Systemic barriers include a lack of IHC expertise and/or a reflex IHC process, a lack of process for the disclosure of results by treating providers, a lack of clear language or directives in the pathology report, a delay between IHC results and the cancer diagnosis, and the physical distance to genetic counseling centers. …There are care provider–related barriers as well because they may not be aware of the importance of genetic assessment for their patients or lack knowledge about logistical details for the coordination of referrals. Adding to this, the workup of LS is molecularly complex because multiple genes can be involved through different mechanisms, and it sometimes requires somatic testing, which

necessitates more guidance for the unfamiliar clinicians. Notwithstanding these barriers, once patients get to their genetic counseling appointment, the uptake of genetic testing is high (77%-90%)'[14].
Education addressing lack of knowledge about the importance of genetic testing is relevant here, but needs to be part of a larger effort to coordinate services and improve communication between non-genetic and genetic healthcare providers to optimise follow-up care and allow for prevention.

Context and limits of education- Sharing and coordinating responsibilities for genetic testing

While education for healthcare professionals is crucial, it alone is insufficient for effective integration of genetic testing into mainstream care. Education must be part of a broader framework that includes clear communication and accessible genetic specialist services. For instance, in the Netherlands recent guidance on requesting genetic testing by non-genetic healthcare providers stipulates that the healthcare provider needs to be in touch with medical genetic professionals or have access to a multidisciplinary team at any time to discuss questions and uncertainties [15]. Effective integration of genomics in healthcare requires clear agreements on responsibilities among clinical staff. A recent communication argues reflection on responsibilities needs to encompass the entire process of genetic testing, and, for instance, include ethical reflection on the effects genetic information may have on patients[16]. For instance, in some cases genetic information may be uncertain and the consequences of the genetic test may not lead to straightforward interventions. 'Ethical reflection should start prior to genetic testing, as findings, even when uncertain, can have important ramifications for the patient…To foster professional collective responsibility in the mainstreaming of genomic medicine, we suggest educating and integrating non-specialists more explicitly throughout all stages of the testing process. Importantly, genetic education needs to be seen as a continuous process rather than a one-time event and should include ongoing interactions with genetic services'[16]. The importance of having open communication channels and clarity on responsibilities is further exemplified by the finding that communication decisions may be impacted by awareness of downstream handling of information. 'Clinical laboratory scientists might report less to nonspecialists than to genetic/genomic specialists, as they feel responsible for minimising the potential of non-specialists misunderstanding, misinterpreting or miscommunicating the genetic findings to patients [16, 17].

Clinical geneticists and their professional and medical organisations, such as the European Society of Human Genetics (ESHG) and national clinical genetics societies should take a leadership role not only in genetic education, but also in agenda setting regarding the coordination of roles and responsibilities in the process of increasing mainstreaming of genetic services in health care [5, 18]. ESHG has an Education Committee that traditionally focussed on genetic professionals but has recognised an 'additional responsibility to provide resources to health professionals and scientists working outside of genomics'[5]. This organisation's

website hosts a link to extensive educational resources [ESHG Genetic Educational Materials & Sources - EuroGEMS.org Home page](EuroGEMS.org Home page).

3 Example of primary prevention

As illustrated by the examples of autosomal dominant disorders mentioned above, secondary prevention for index cancer patients may involve treatment adaptations such as more elaborate surgery. For family members, genetic testing can guide primary or secondary prevention, such as removal of specific tissue, monitoring through mammography or MRI (Magnetic Resonance Imaging), for breast cancer, or colonoscopy for colorectal cancer. In case of cardiovascular disorders, cardiological surveillance may be started, or medication, such as statins may be prescribed in case of Familial Hypercholesterolaemia (FH) to prevent or delay the onset of coronary artery disease.

In a few countries, e.g. the Netherlands and Norway, screening programmes for FH have been organised since the 1990s to allow for a more systematic, public health approach towards identifying and informing family members for genetic testing and prevention [19]. Such an approach is relevant as the autosomal dominant form of FH is relatively frequent with a prevalence of 1:200 - 1:500 worldwide. FH is caused by mutations affecting cholesterol metabolism or lipoprotein receptors and effective medication is available in the form of statins [20]. Detection can be based on high cholesterol, or via genetic screening. The Dutch screening programme has ended and FH screening is now integrated in regular health care. Coordination of identifying FH patients and support for informing the family is delegated to the LEEFH foundation (National Expertise Centre for Genetic Testing for Familial Cardiovascular Diseases).

LEEFH offers targeted education and training for healthcare professionals involved in FH care. Through the LEEFH platform, GPs and other healthcare providers can access up-to-date resources and training modules that enhance their ability to identify, manage, and treat patients with FH effectively. This approach hinges on general practitioners (GPs) who in the Netherlands are able to request genetic testing for FH themselves. Not all GPs are aware of this possibility. LEEFH informs about this option and about the procedure for drawing blood and requesting the genetic testing. This organisation also dispenses the testing kits and is in close contact with internal medicine specialists and the lab testing the mutations. Clinical geneticists are not routinely involved, though they may be in some countries. Internal medicine physicians or vascular medicine specialists may have extensive knowledge on the genetic mechanisms involved in FH. It is advised to have a referral to an internal medicine

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specialist in a hospital with FH expertise to allow for confirmation of the diagnosis and treatment plan, after which the GP takes on the regular care for the patient. Also in this case the importance of communicating the relevance of informing first-degree family patients members to index patients may not be clear for GPs or regular vascular medicine specialists who are used to focussing on individual patients. Having a dedicated organisation that is able to coordinate education and coordination of care and prevention supports the broader goal of integrating genetic knowledge into everyday clinical practice for improved personalised prevention [19]. Also in this case having access to resources and expert knowledge supports mainstream care professionals in providing adequate care and enabling prevention.

4 Example of secondary prevention

In prescribing medication, pharmacogenetic information may be used to improve safety and effectiveness and avoid adverse drug reactions. Widely-used guidelines have been developed by the Clinical Pharmacogenetics Implementation Consortium (CPIC) and the Dutch Pharmacogenetics Working Group (DPWG) [21]. Pharmacogenetic testing may be done prior to presciption (preemptive testing), at the time of prescription (companion testing) or after a prescribed drug fails to have an effect or has adverse effects (reactive testing). Pharmacogenetic expertise may be located in various hospital departments, such as clinical pharmacy or medical genetics, but also mainstream healthcare providers and medical specialists such as psychiatrists and oncologists may be applying pharmacogenetic information. In primary care GPs and pharmacists may prescribe medication for which pharmacogenomic guidance is available. Many studies underscore the lack of education as an important barrier to implementation of pharmacogenetics [22-24].

Several strategies are reported to support healthcare providers in applying pharmacogenetic information, or to boost their pharmacogenetic knowledge. One way to support clinicians in prescribing medication is by using information and communication technology (ICT). Clinical decision support (CDS) systems, connected to electronic health records, may be used to alert the physician when prescribing medication via an active pop-up about the availability of pharmacogenomic guidance on that medication (pre-test alert) or issue a recommendation after a high-risk pharmacogenomic test result becomes available (post-test alert). Such pop-ups can be described as real-time education or just-in-time education [24]. To increase and improve pharmacogenetic knowledge of healthcare providers just-in-time education can be complemented with access to resources such as guidelines from CPIC or DPWG [16].

Another promising strategy to improve knowledge of both clinicians and clinical pharmacists is by promotiong interprofessional education, most notably among pharmacology and medical students. For instance by bringing these students together in person or via tele-health systems to work on specific cases starting from their own expertise stimulating the exchange of

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perspective [25].

5 Example of tertiary prevention

Especially in oncology, treatment-related genetic information is increasingly being incorporated into care pathways, as knowledge on tumour characteristics and targeted therapies can be used for tertiary prevention, aiming to reduce or eliminate the progression of the disease. Skills needed include knowledge on the availability of tumour testing, and knowledge on guidelines on how and when to request such testing and how to implement the test results in treatment decisions, affecting roles and responsibilities for professionals involved in this care [26, 27]. A recent international survey among hematologists and hemato-oncologists in France, Germany and the United States identified four areas of educational needs: '(1) sub-optimal knowledge of treatment guidelines; (2) sub-optimal knowledge of molecular testing to inform…treatment decisions; (3) sub-optimal skills when making treatment decisions according to patient profile (co-morbidities, molecular testing results); and (4) challenges balancing the risk of toxicities with benefits of treatment . Over one-third of the respondents reported skill gaps when selecting suitable treatment options and prescribing therapies and reported a lack in confidence to initiate and manage treatment [28].' The authors suggest a need for 'continuing medical education specifically to improve knowledge of treatment guidelines, and to assist clinicians in developing skills and confidence when faced with clinical decision-making scenarios of patients with specific comorbidities and/or molecular test results, for example, through case-based learning activities' [28].
In addition to clinicians, also other health care professionals, such as nurses are in need of training to better understand and support this highly specialised form of care and help communicate aspects of this care to patients [26, 29-31].

6. Points to consider for developing guidelines on genetic education for non-genetic healthcare professionals in personalised prevention

- Inclusion of genetic education in medical curricula:

  Genetic education should be a core part of all medical training. Healthcare professionals need such a foundation to understand the basics of the genetic architecture of disorders, be able to assess patients for referral to clinical genetics centres, communicate about test results, understand the impact of test results on their patients, and also, under specific circumstances, be able to request genetic testing themselves.

- Ongoing genetic education, also during medical specialisation training:

  Continued genetic education should be used to build on and expand knowledge and understanding of applying genetic information in health care, especially when mainstreaming of genetic services are becoming part of healthcare professional's tasks. Education should include awareness also of ethical aspects, impact of genetic information and potential uncertain information on patients and the wider implications for family members.

- Access to genetic expertise:

  Although education is a key element in fostering the ongoing integration of genomic information in health care and prevention, education alone is not enough. Access to genetic expertise is vital to support health care professionals in decision making regarding requesting testing, communicating about test results and incorporation genetic information into treatment and prevention plans.

- Clear Protocols:

  Clear protocols are vital to define roles and responsibilities for each step of a care pathway that involves genetic information. Collaboration to establish responsibilities and education regarding tasks needs attention at an institutional, national and international level, in which professional and medical organisations of geneticists and other medical specialties can play a key role.

- Information and Communication Technology:

  ICT systems are helpful to support clinical decision making based on patient characteristics and available information on protocols for e.g. pharmacogenetic testing, and can support just-in-time education on applying genetic information fostering the integration of personalised prevention in healthcare.

1. Nisselle, A., et al., *Ensuring best practice in genomics education: A theory- and empirically informed evaluation framework.* Am J Hum Genet, 2024. **111**(8): p. 1497-1507.
2. George, A., et al., *Implementing rapid, robust, cost-effective, patient-centred, routine genetic testing in ovarian cancer patients.* Sci Rep, 2016. **6**: p. 29506.
3. Eikenboom, E.L., et al., *Universal Immunohistochemistry for Lynch Syndrome: A Systematic Review and Meta-analysis of 58,580 Colorectal Carcinomas.* Clin Gastroenterol Hepatol, 2022. **20**(3): p. e496-e507.
4. EBMG. *European Board of Medical Genetics*. 30 August 2024]; Available from: https://www.ebmg.eu/413.0.html.
5. Tobias, E.S., et al., *The Role of the European Society of Human Genetics in Delivering Genomic Education.* Front Genet, 2021. **12**: p. 693952.
6. Hilary Burton, A.H., Mark Kroesen, Sobia Raza, *Genomics in mainstream clinical pathways*. 2017, PHG Foundation.
7. Monaghan, A., E. Copson, and R. Cutress, *Hereditary genetic testing and mainstreaming: a guide for surgeons.* Ann R Coll Surg Engl, 2024. **106**(4): p. 300-304.
8. Cormack, M., et al., *Mainstreaming genomic testing: pre-test counselling and informed consent.* Med J Aust, 2024. **220**(8): p. 403-406.
9. Bunnik, E.M., et al., *Mainstreaming informed consent for genomic sequencing: A call for action.* Eur J Cancer, 2021. **148**: p. 405-410.
10. Ormond, K.E., et al., *Defining the Critical Components of Informed Consent for Genetic Testing.* J Pers Med, 2021. **11**(12).
11. Ahsan, M.D., et al., *Do people with hereditary cancer syndromes inform their at-risk relatives? A systematic review and meta-analysis.* PEC Innov, 2023. **2**: p. 100138.
12. Menko, F.H., et al., *The uptake of presymptomatic genetic testing in hereditary breast-ovarian cancer and Lynch syndrome: a systematic review of the literature and implications for clinical practice.* Fam Cancer, 2019. **18**(1): p. 127-135.
13. Cragun, D., et al., *Comparing universal Lynch syndrome tumor-screening programs to evaluate associations between implementation strategies and patient follow-through.* Genet Med, 2014. **16**(10): p. 773-82.
14. Kim, S.R., et al., *Maximizing cancer prevention through genetic navigation for Lynch syndrome detection in women with newly diagnosed endometrial and nonserous/nonmucinous epithelial ovarian cancer.* Cancer, 2021. **127**(17): p. 3082-3091.
15. Netherlands, V.S.C.G.i.t. 2024 22 August 2024]; Available from: https://www.vkgn.org/nieuws/leidraad-aanvragen-genetische-kiembaandiagnostiek/.
16. Siermann, M., et al., *Stimulating professional collective responsibility from the outset in mainstreaming genomics.* J Med Ethics, 2024. **50**(8): p. 525-526.
17. Sahan, K., et al., *Ethical preparedness in genomic medicine: how NHS clinical scientists navigate ethical issues.* J Med Ethics, 2024. **50**(8): p. 517-522.
18. B1MG, *Policy Brief on implementing Genomics in Healthcare*.
19. Leren, T.P. and M.P. Bogsrud, *Cascade screening for familial hypercholesterolemia should be organized at a national level.* Curr Opin Lipidol, 2022. **33**(4): p. 231-236.
20. Louter, L., J. Defesche, and J. Roeters van Lennep, *Cascade screening for familial hypercholesterolemia: Practical consequences.* Atheroscler Suppl, 2017. **30**: p. 77-85.

21. Bank, P.C.D., et al., *Comparison of the Guidelines of the Clinical Pharmacogenetics Implementation Consortium and the Dutch Pharmacogenetics Working Group.* Clin Pharmacol Ther, 2018. **103**(4): p. 599-618.
22. Just, K.S., et al., *Medical education in pharmacogenomics-results from a survey on pharmacogenetic knowledge in healthcare professionals within the European pharmacogenomics clinical implementation project Ubiquitous Pharmacogenomics (U-PGx).* Eur J Clin Pharmacol, 2017. **73**(10): p. 1247-1252.
23. Rigter, T., et al., *Implementation of Pharmacogenetics in Primary Care: A Multi-Stakeholder Perspective.* Front Genet, 2020. **11**: p. 10.
24. Haidar, C.E., et al., *Advancing Pharmacogenomics from Single-Gene to Preemptive Testing.* Annu Rev Genomics Hum Genet, 2022. **23**: p. 449-473.
25. Lee, Y.M., et al., *Advancing Pharmacogenomics-Based Care Through Interprofessional Education.* Am J Pharm Educ, 2023. **87**(5): p. 100007.
26. White, S., C. Jacobs, and J. Phillips, *Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care.* Genet Med, 2020. **22**(7): p. 1149-1155.
27. Daly, R., et al., *Precision Medicine Is Changing the Roles of Healthcare Professionals, Scientists, and Research Staff: Learnings from a Childhood Cancer Precision Medicine Trial.* Journal of Personalized Medicine, 2023. **13**(7): p. 1033.
28. Peloquin, S., et al., *Knowledge, skills, and confidence gaps impacting treatment decision making in relapsed/refractory chronic lymphocytic leukemia and mantle cell lymphoma: a quantitative survey study in France, Germany, and the United States.* BMC Cancer, 2024. **24**(1): p. 1003.
29. Laaksonen, M., E. Airikkala, and A. Halkoaho, *The Development of Education of Public Health Nurses for Applying Genomics in Preventive Health Care.* Front Genet, 2022. **13**: p. 849232.
30. Rahman, B., et al., *Genetic and genomic learning needs of oncologists and oncology nurses in the era of precision medicine: a scoping review.* Per Med, 2022. **19**(2): p. 139-153.
31. Diez de los Rios de la Serna, C., P. Fernández-Ortega, and T. Lluch-Canut, *Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study.* Journal of Personalized Medicine, 2022. **12**(7): p. 1104.

D5.1.C Communication to family members

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Table of content

Contents

Table of Acronyms

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CSA: Coordination and Support Action

ELSI: Ethical, legal and societal implications

WP: Work package

KUL: KU Leuven

VUMC: Vrije Universiteit Medisch Centrum (Amsterdam UMC)

EPF: European Patients' Forum

ACN: Active Citizenship Network

D5.1.C Communication to family members

This section of the deliverable addresses the work carried out in task 5.2 'Definition of guidelines for communication to family members' (KUL, VUMC, EPF; ACN). In this task, we examined issues related to communication of genomic findings with familial implications by critically analyzing ethical, legal and social implications (ELSI) literature. This analysis led to the creation of a draft document containing recommendations for the return and communication of results within the context of Personalized Prevention.

On June 19-20, 2023, a two-day workshop was held in the historical city of Leuven (Belgium). The workshop brought together an expert panel of inter-disciplinary stakeholders who are leading work in the area of disclosure of genetic information to family members through research, clinical practice or policy. The primary aim of this workshop was to support the further development of guidelines for returning genomic test results with familial consequences in the context of personalized prevention. The expert panel discussed various approaches currently used in the UK, France, Denmark and Australia. Furthermore, in-depth discussions were held on the responsibilities of patients and healthcare professionals, as well as the potential for applying a public health approach. These discussions were informed by the ELSI literature review that had been conducted.

An agenda setting paper is currently under preparation in collaboration with the workshop participants and will be submitted to Open Research Europe for peer review upon completion.

Introduction

The field of genetic and genomic medicine, recognized for its significant potential to advance personalized medicine, presents substantial opportunities for the precise tailoring of healthcare interventions based on individual genetic profiles. Genomic sequencing is being increasingly implemented into mainstream clinical practice. Results, which can sometimes be unexpected, can have implications not only for individuals, but also for their relatives. And while genetic information may be considered personal, it is also inherently familial (Lucassen 2007). Identification or confirmation of a hereditary condition (whether by DNA analysis or another method) in an individual can indicate that other family members may also develop the condition and could also pass it to any future children. When a heritable genetic condition is present in a family, disseminating that information to family members plays a vital role in giving them the option to undergo early testing, prevention, and (potentially) treatment. Furthermore, information about heritable genetic risk can enable more informed reproductive decision making and may be used to prevent passing the condition on to the next generation.

Although the familial communication of genetic information is widely seen as important, discussing genetic risk in a family can cause distress. Some patients struggle to initiate these conversations with their family members (Gaff et al., 2007). Conversations can be challenging, and patients may worry that disclosing information about genetic risk could be harmful to their relatives. It can be difficult for patients to find the “right time” to broach the subject (Derbez 2018). In some cases, patients may feel guilty about the genetic diagnosis. Furthermore, patients may feel overly burdened with the responsibility to inform their relatives, especially considering that some may be coping with their own diagnosis and treatment at the time. All of these concerns are further compounded when the patients have distant or strained relationships with their relatives (Black et al., 2013; Clarke et al., 2005; Geelen et al., 2011; Suthers et al., 2006; van den Heuvel et al., 2019). So, despite the fact that most patients have the intention to inform their at-risk relatives, empirical research shows that in practice this communication does not always occur (Menko et al., 2013; Pedrazzani et al., 2022, Daly et al., 2016).

Knowing the challenges that patients might face with familial communication of genetic risk, healthcare professionals could have mechanisms to support patients in the process. The primary way in which they do this is by offering a ‘family letter’ which informs the recipients about their genetic risk and encourages them to seek testing (Dheensa et al., 2018). Usually, the patient is given the letter and asked to distribute it to relevant family members. However, in some cases, with the patient’s consent, the healthcare professional may distribute it on their behalf. If available, additional counselling may also be provided to patients who need further support with informing their relatives.

Despite the availability of support to assist family communication, a proportion of patients still do not communicate genetic risk information to family members. In this instance, the patient’s right to confidentiality and autonomy comes in conflict with the interests of the relatives who could benefit from knowledge of a genetic risk in the family. In cases where the healthcare professional is aware of nondisclosure, they are confronted with the dilemma of how to balance these two conflicting sides; while they have a duty to care for their patient and a duty to protect their confidentiality, they may also feel responsible, or might in fact be responsible, for preventing harm for the patient’s at-risk relatives. While not common, during

the course of their careers many healthcare professionals encounter this dilemma yet feel that they lack the proper resources to help them make a decision (Clarke et al., 2005; d'Audiffret Van Haecke and de Montgolfier, 2016).

Although the patient has a legal right to confidentiality, this is not absolute, with exceptions already being widely accepted in the context of infectious diseases or when a patient poses a serious and imminent threat to themselves or others. The question arises whether, in the context of genetics, a breach in confidentiality can ever be warranted and if so, under which conditions. Alternatively, some authors sidestep this dilemma by positing that disclosing genetic risk information to family members does not necessarily violate the patient's confidentiality (Parker and Lucassen, 2004; Lucassen and Gilbar, 2018; Gilbar and Barnoy, 2020). The crux of this argument rests upon the fact that while in some ways genetic information is personal, it is also inherently familial and shared. This means that confidentiality can be considered at a familial level whereby genetic information belongs to both the individual patient and their family (Lucassen 2007). Informing relatives of a genetic risk in the family thus does not breach the patient's confidentiality because as Lucassen and Gilbar (2018) argue "no identifiable information is communicated in such a statement, even if genetic findings in one person first led to that conclusion."

To add a further layer of complexity to the ethical challenge of cases of nondisclosure, there is also the relative's right to know and right not to know information about their own genetic risk. On the one hand, relatives may have a legal right to information that pertains to their health and that could prevent harm (Brownsword and Wale, 2017). On the other hand, relatives may have a right not to know information that they might find distressing or irrelevant either clinically or regarding their everyday life (Mendes et al., 2019). Notably, some argue that informing someone that they are at risk does not necessarily violate the right not to know since the person informed can still choose whether or not they want to get tested, but this argument remains controversial. In several jurisdictions the right to know and the right not to know are not only ethical rights, but also have a legal backing (Brownsword and Wale, 2017). Whoever is disclosing the information, whether it is the patient or the healthcare professional, must consider these possibly competing rights when making decisions about disclosure.

On June 19th and 20th 2023, a workshop was held in Leuven, Belgium with an expert panel consisting of interdisciplinary stakeholders (see annex for full panel composition) involved in the area of family communication of genetic risk information either through research, clinical practice, or policy. Participants came from eight countries, namely Belgium, the Netherlands, France, the UK, Australia, Denmark, Sweden, and Portugal. Organized in the context of the Horizon Europe project PROPHET (PeRsOnalised Prevention roadmap for the future HEalThcare in Europe), the workshop aimed to support the development of guidance for returning genetic test results with familial consequences in the context of personalized prevention. The workshop consisted of a general overview of the ethical issues involved in family communication, as well as presentations on specific policy approaches adopted by France, Australia, the UK, and Denmark. These concrete examples provided reference points for the ensuing in-depth discussions on the roles and responsibilities of patients and healthcare professionals, and whether a public health approach should be applied to ensure at-risk individuals are informed. These discussions explored different areas of personalized prevention, including clinical genetic care and public health screening programs.

Based on the input gained during the workshop discussions, this paper provides an overview of the ethical and legal landscape as well as identifying future research priorities. From the discussions five main themes emerged that we hope will foster conversation on the topic, namely: 1) the emphasis on family communication as a process, 2) the shift to a familial approach, 3) roles and responsibilities in the communication process, 4) the need to clarify guidelines and policy, and 5) the importance of adequate resources.

Emphasis on family communication as a process

As a whole, participants emphasized how family communication is a complex, multi-step process. Participants pointed out how the language of disclosure currently used in the clinic, literature, guidelines, and policies can imply that family communication is a one-time event rather than an ongoing process in need of continual re-evaluation and revisitation. To enhance this process, several participants proposed that prior to testing, healthcare professionals and patients create an informal "contract" between both parties to clarify respective responsibilities and expectations to prevent misunderstandings following the return of results.

The complexity of the process of family communication necessitates a nuanced approach by the healthcare team to support patients along this journey, creating a shared responsibility between healthcare professionals and patients. Participants noted the importance of distinguishing between merely transmitting information and engaging in meaningful family communication, requiring healthcare professionals to encourage and support this dialogue beyond information provision and also beyond the consultation where the results are returned. Participants advocated for patient involvement from the outset of genetic testing, with an emphasis on informing them about the possible consequences of genetic tests and addressing their concerns. Clear communication guidelines, including who, what, why, and when to inform, supported by communication tools, can assist both patients and healthcare professionals in navigating expectations and responsibilities throughout the process of family communication. These can then be adapted to suit the contextual features of different patient and clinical scenarios, as well as the potential utility of the information.

The importance of healthcare professionals and patients discussing familial implications and dissemination of results together throughout the genetic testing or genomic sequencing timeline (i.e., during pre-test counselling, return of results, and follow up) emerged as a core theme of the workshop discussions as we discuss below in further detail.

Pre-test counselling

Research indicates that addressing family communication earlier in the genetic testing or genomic sequencing process improves rates of disclosure in families (Young et al., 2019; Pedrazzani et al., 2022). However, evidence also suggests that the possibility of uncovering results with familial implications is not uniformly addressed in pre-test consultations. In some cases, familial implications are discussed prior to testing since they are the main aim of testing, but in other instances, familial implications are seen as more of a secondary outcome and thus not addressed at all pre-test. For instance, when a patient has colorectal cancer, they might be encouraged to get genetic testing in order to help their relatives by determining whether there is a pathogenic variant in the family. This contrasts with cases where patients present with symptoms with no known cause, and thus finding a diagnosis for the patient is the primary objective of testing meaning that familial implications may or may not be addressed before testing.

While participants of the workshop generally agreed that discussing family communication prior to sequencing is best practice, some pointed out how, in some contexts, adhering to this standard may prove challenging. For example, when genomic sequencing, as opposed to targeted genetic testing, is used, due to its broader scope, it is more likely to generate unexpected and uncertain findings. The difficulty with predicting findings in this context understandably complicates the clinical team's ability to inform patients about the possibility for familial implications. Participants raised that informing patients of this potential outcome without being able to provide further information about the kind of genetic risk that could be identified could be potentially distressing for patients. Bearing in mind these challenges, participants nevertheless generally agreed that, to the best of their abilities, healthcare professionals should discuss the potential for familial implications prior to testing to help make patients aware of this possibility and the need to inform their relatives. Furthermore, one participant noted how initiating conversations with family members prior to testing could in some cases lower the barriers for patients to eventually share their results.

Post-test consultation

The expert panel agreed that when healthcare professionals return results, it is important that they also address familial implications and the importance of disseminating information about the genetic risk within the family. Healthcare professionals should help patients to identify which family members are at-risk and could benefit from being informed. At the same time healthcare professionals should ensure that patients understand that a heightened risk is not the same as a diagnosis. While this was noted by participants to be standard practice in their respective contexts (e.g. clinical genetic services), empirical research with patients demonstrates that patients often misunderstand or do not sufficiently understand who should be informed and what to communicate to relatives (Dheensa et al., 2018; Menko et al., 2013; Mendes et al., 2019). Participants of the workshop supported current practice whereby in the post-test consultation healthcare professionals provide patients with family letters to help them disseminate information about the familial genetic risk. If it appears that a patient will not inform their relatives, participants identified connecting patients with additional counselling or psychological resources as an important strategy to support patients.

Follow up

Workshop attendees emphasized the importance of following up with patients after the initial return of results as a crucial step in ensuring disclosure. Those working in clinical practice supported the research finding that most patients do not outwardly object to informing their relatives (Gilbar and Barnoy, 2020). Instead, many patients intend to communicate, but postpone doing so due to uncertainty about how to approach the subject and concerns about potentially causing distress to their relatives (Pedrazzani et al., 2022; Li et al., 2018; van den Heuvel et al., 2019). Patients may struggle to process that others in their family are at-risk and may even feel guilty about informing their relatives, especially if they feel responsible for passing the condition on to their children. At the time of the initial return of results some patients may be overwhelmed by the additional burden of having to inform their relatives since they may already be struggling to cope with their own diagnosis and treatment. In light of these challenges, workshop discussions emphasized the value of following up with patients about family communication after some time has elapsed. If a patient appears to be struggling with informing their relatives, a follow up meeting can be an important way to ensure that they are provided with additional support to help them communicate. Participants also identified genetic counsellors, psychologists, and general practitioners as key potential actors

in the follow up process. While participants supported the development of follow up procedures, they noted how the lack of dedicated resources may be an obstacle to implementation and advocated that sufficient resources be made available.

Shift to a familial approach

Throughout the workshop discussions, there were several times in which tensions arose between policies focused on individual rights compared with the perceived familial and relational nature of genetic information. Critically examining the individualistic tendencies in our ethical and legal systems, the discussions highlighted the imperative to adopt a more relational approach in clinical practice. This potentially paradigmatic shift away from an exclusive focus on individual rights and responsibilities would require a reconsideration of the patient's rights framework, to emphasize the inherent familial nature of genetic information and steer away from an exclusive focus on the individual.

Advocating for a broader perspective in clinical genetic practice, the proposal discussed by participants was to treat the family as the primary unit of care, urging that ethical guidelines and policies align with this familial and relational approach. This approach has also been advocated for in the literature, most notably by Lucassen (2007), whose joint account model acknowledges the familial aspect of genetic information obtained from an individual (index patient/proband) and underscores the possibility of disclosing such familial information without breaching patient confidentiality. However, participants noted that if the unit of care does shift beyond the patient, there arises a need for a clearer definition of "at-risk relatives."

While participants generally supported this shift in theory, several raised concerns about its translation into practice. Participants noted how despite the increasing traction in the ethical literature, implementation has been much less straightforward in countries that have adopted familial and relational language in their guidelines and policies on genetic risk communication. For some these challenges in implementation made this shift seem like a distant possibility, with many legal reforms being required to make this reframing tenable. For other participants, there was a desire for an aspirational push to reform legal framing to indicate that the use and disclosure of familial information for the care of relatives should be deemed a reasonable expectation for clinical genetic services, while concurrently supporting and encouraging the index patient as a matter of good practice.

Roles and responsibilities in the communication process

In some circumstances it was agreed that patients are best situated to inform at-risk relatives due to their (presumably) pre-existing relationship, with one participant even noting how the act of communicating genetic risk can help strengthen or reinforce relationships. However, another central theme underpinning many of the workshop discussions was an acknowledgement of the limitations of patient led (also known as proband-mediated or family-mediated) communication. This aligns with empirical research that indicates that, at least for some conditions, a significant portion of at-risk relatives may not be sufficiently informed of their genetic risk. In light of these challenges, participants expressed the need to explore alternative approaches, whereby healthcare professionals adopt a more active role in the communication process. Alternatives to patient-led disclosure have been gaining traction, with countries such as the Netherlands, Switzerland, Sweden, Denmark, Finland, and Australia, investigating the acceptability of different levels of involvement of healthcare professionals in recent years (Pedrazzani et al., 2022; Menko et al., 2013; Andersson et al.,

2020; Hawranek et al., 2021; Petersen et al., 2019). While there has been a lot of scepticism towards moving away from a purely patient-led approach to disclosure, an increasing amount of literature shows that patients and the public may not be averse to disclosure led by healthcare professionals or healthcare services (Andersson et al., 2020; Hawranek et al., 2021; Petersen et al., 2019).

The workshop did not reach a clear consensus on the degree of involvement that healthcare professionals should have in the communication process, and opinions were particularly divided regarding cases of patients explicitly objecting to disclosure (active nondisclosure). Some were supportive of healthcare professionals having the ability to inform at-risk relatives (especially those from legal systems from which this is already possible) due to the potential benefit to the patient's relatives. Others were sceptical due to concerns over the patient-clinician relationship (trust, assumption of care), and concerns that this will be overly burdensome to healthcare professionals. Although there are some concerns with a direct contact approach to disclosure whereby healthcare professionals rather than patients inform at-risk relatives, it is not clear that these limitations outweigh the limitations of patient led communication. Participants noted that direct contact can be an appealing alternative in cases where the patient has a distant or strained relationship with relatives. Exploring whether the patient's clinical geneticist is the best person to lead disclosure or whether there could be an added benefit to communication led by general practitioners, the patients' treating specialist or public health authorities was identified as an area for further research.

Exploring the role of non-genetics healthcare professionals in disclosure

Workshop discussions raised the question of whether practitioners outside of the genetics specialty could be better situated to facilitate follow-up with patients and disclosure to their at-risk relatives. For instance, one participant suggested that primary care providers try to follow up with patients to see whether they have informed their relatives or whether they need further support. Expanded collaboration between genetics specialists and other healthcare professionals could help redistribute some of the responsibility and resource burden placed on genetics centres. Furthermore, structural patient engagement could help to develop new strategies. Several participants noted that as genetics and genomics become increasingly mainstream, healthcare professionals such as oncologists, cardiologists, pediatricians, neurologists and general practitioners have an increasingly important role in providing genetic testing. It is therefore critical that future studies explore the experiences, attitudes, and practices of other healthcare professionals outside of the genetics specialty, as the potential role in family communication of other health care professionals remains underexplored. Another suggestion was that it may be advantageous to inform relatives using a third party with an established relationship with the relative. This could help to reduce the conflict that healthcare professionals might experience if they are to inform relatives, particularly if it is against patient wishes. Currently, standard practice for contacting at-risk relatives is through family letters, but informing relatives through another healthcare professional could help to link relatives to the healthcare system which can help support them to make a decision about pursuing counselling or testing.

Guidelines and policy

Some countries have policies that specifically address the issue of family communication of genetic risks, although the approaches taken differ significantly. In Australia, for example, regulations made under Federal legislation (and mirrored in some states) have made it

possible for some healthcare professionals to breach confidentiality to inform family members in some circumstances (Otlowski 2013; Otlowski 2015; Australian Privacy Act 2019). In the UK, the recent court case ABC v St Georges Healthcare NHS Trust & Ors takes the Australian regulations a step further by asserting that genetic healthcare professionals have a legal duty to weigh the interests of patients and their at-risk relatives, meaning in some exceptional cases a healthcare professional could be obliged to inform relatives of their genetic risk (Middleton et al., 2020). In the US, the court case Safer v. Estate of Pack (1996) established a similar precedent whereby healthcare professionals had a duty to warn at-risk relatives that could not always be sufficiently satisfied by only warning the patient. However, genetic privacy statutes and the later enactment of the Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule effectively overruled the previous case law, meaning that healthcare professionals are neither required nor permitted to disclose without patient consent (Rothstein 2018). In France, a law has imposed a duty on the patient to inform family members either directly or indirectly via their healthcare professional (French Bioethics Law 2011). More commonly, as is the case in Belgium, no specific guideline or law exists that addresses what can be done in cases of nondisclosure (Phillips et al., 2022).

One of the primary goals of organizing the workshop was to stimulate discussion on the advantages and challenges of various proposed policy approaches for communicating genomic findings with familial implications. This dialogue aligns with PROPHET's broader objectives of helping public health authorities enhance their ability to design, assess, and implement effective personalized preventive approaches. By exploring diverse policy perspectives, the workshop provides valuable insights that can inform and guide the further development of improved criteria/guidelines for returning and communication genomic results with familial implications in the context of personalized prevention.

While the standard approach in all countries represented in the workshop was that disclosure of the result of a genetic test or information on a familial risk is only permitted with the consent of the patient, in some there was an exception to patient confidentiality (at least in its current individualistic formulation) granted when the genetic condition in question is both severe and actionable (Phillips et al., 2021). Despite severity and actionability being commonly used criteria between various national contexts, workshop participants pointed out the challenges of defining these boundaries given the variable penetrance, and expression of genetic conditions which is inherently probabilistic. While a few participants floated the idea of having a fixed list of conditions to help decision aids regarding familial disclosure, others were quick to point out how this approach may be overly restrictive and not allow for enough leeway to consider case particulars, such as the nature of the genetic information and the familial situations. Using criteria instead can help allow for a wider set of factors to be taken into account. However, as a consequence of not having a fixed list, it can be difficult to determine where exactly to draw the line between when it is (and is not) acceptable to disclose genetic risk information to at-risk relatives without the consent of the patient.

While there was a lack of consensus among participants regarding which policy approach best balanced the interests of patients, at-risk relatives, and healthcare professionals, participants did agree that at the very least clear guidelines and legislation are needed to help mitigate confusion and conflict in this ethically complex situation. In countries where the legislation was ambiguous and no guidelines were present, legislation may only be applicable by analogy, meaning more than one policy approach could be possible. Participants noted that misunderstandings occurred and that healthcare professionals experienced distress over

having no clear guidance on what would be considered best practice in this scenario. With no clear guidance, healthcare professionals are left to make decisions on their own, which one participant stated could cause inconsistencies in care between providers. Workshop discussions made it clear that regardless of which policy was adopted, the legal parameters as well as organizational and resource availability would need to be compatible with the approach for the change to be tenable in practice. Rather than endorsing a single policy, some participants opined that a combination of approaches may be most appropriate given the different needs and preferences of patients and the wide range of genetic conditions.

Participants in countries where a specific policy regarding this issue had been adopted were quick to point out the challenges regarding interpretation and implementation. Policies remain far from straightforward, and healthcare professionals still encounter uncertainty in how to handle ethically sensitive cases where patients are unable or unwilling to communicate with their at-risk relatives. These findings from the workshop have similarly been reflected in the literature originating from the UK, France, and Australia (D'Audiffret Van Haecke and de Montgolfier, 2016; D'audiffret Van Haecke and de Montgolfier 2018; Dheensa et al., 2017; Meggiolaro et al., 2020). Participants seemed to agree that continual investigation and evaluation of guidelines and policies is needed to improve their implementation and best mitigate their drawbacks.

Below we present the three key policy areas discussed during the workshop:

The patient's role in disclosure

A consensus was established early in the workshop that patients have an ethical responsibility to inform their at-risk relatives. Participants were in agreement that healthcare professionals should emphasize this responsibility to patients at various time points and proactively offer support. The point of divergence in opinions was whether this ethical responsibility should be supported by legislation. Many participants expressed concerns about whether such a framework might impose legal consequences on patients who fail to inform their relatives. As of yet, in France the effects of the legislation have been seemingly less direct, with French participants noting how their national legislation helps emphasize to patients and healthcare professionals the importance of informing at-risk relatives. This sentiment has been echoed in empirical research conducted with French genetics professionals who when asked about the law's impact on practice stated that in cases where patients appeared hesitant to disclose, the weight of the law could be a helpful tool to implore patients to inform their relatives (D'Audiffret Van Haecke and de Montgolfier 2016). Despite these purported benefits, several participants remained unconvinced that the patient's ethical duty should translate to a legal duty. These participants were concerned that if legislation results in lawsuits being filed against family members for a failure to communicate, this could be very damaging and in conflict with the initial aim to prevent harm. Another participant also cautioned against using cases of active nondisclosure, which are an outlier, as the basis for legislation stating instead that proposed policies must be proportionate and not lose sight of the much more common type of nondisclosure (passive nondisclosure) where patients do want to share relevant information with their families but may have misunderstood what to disclose or to whom the information is relevant and just need more support.

Healthcare professionals' roles and responsibilities

While many workshop participants agreed that healthcare professionals have some kind of responsibility towards at-risk relatives, they were divided on their interpretations of what constitutes fulfilment of this duty. Some felt that it was sufficient to inform patients of the

importance of communication with relatives and to provide counselling if requested; while some healthcare professionals may want to do more, these participants felt that it is important that the inherent limitations of this process are acknowledged, particularly since instances of active nondisclosure make up only a small percentage of cases.

In contrast, others were of the opinion that further action is needed to ensure that information about genetic risk is disseminated within the family. There is a need to develop a consensus on what constitutes good practice regarding the healthcare professional's duty. Given the prevalence of (active or passive) nondisclosure and the harm that can occur as a result, it might not be sufficient to just inform patients of the importance of communication. Based on the workshop discussions, we therefore suggest that healthcare professionals be obliged to encourage disclosure and also to follow up with patients about communication. If healthcare professionals suspect that patients have not informed their at-risk relatives, they should again emphasize the importance of disclosure to patients and offer genetic counselling and psychological support to encourage and support disclosure.

A public health approach: the role of national registries

Most participants prior to the workshop were unaware that in some countries national registries have been utilized to contact relatives at-risk of certain genetic conditions. It seemed that this was, in part, because public health discussions have happened in parallel to the bioethical discourse on the topic. The high prevalence of conditions such as hereditary colorectal, breast, and ovarian cancers in the general population and the amount of risk reducing and surveillance options available to reduce the burden to those at risk and on the healthcare system more broadly have been the motivation in some countries adopting this approach. While most participants felt that this approach would not be suitable for all genetic conditions, they did see its value in this specific context and believed this approach could serve well in conjunction with one of the other policy approaches. However, they did note that the implementation of such an approach required significant national infrastructure and public trust in the healthcare system, which in some countries seemed unlikely to be possible in the near future. Efforts would need to be made to ensure that patients and the public are aware of the existence of the possibility of the healthcare system disclosing genetic information to family members and that their ability to choose or opt-out may be limited in this particular context.

Adequate resources

Even if the ethical and legal conflicts on the issue of family communication can be resolved, significant practical challenges remain. To better support patients in the communication process, participants stated that governments must invest in both practitioner training and health infrastructure to ensure necessary resources are available. The practitioners attending the workshop stated several times how they already faced significant time and resource constraints that limit the length and depth of consultation with patients. Currently some healthcare professionals do not have the time available to address familial implications prior to testing. Following testing, healthcare professionals again do not always have sufficient time to address these issues during the post-test consultation, and there is no current systematic support for follow up with patients about family communication. When genetic testing is introduced more broadly in the healthcare system, non-genetic healthcare professionals need education in genetics and either time to support patients in family communication or resources for referral to a clinical genetic unit for counselling of the patient, including support

in risk disclosure to at-risk relatives. Without adequate competence, or time, healthcare professionals cannot be expected to sufficiently address results with familial implications and the importance of family communication. Therefore, structural support to help alleviate restraints on healthcare professionals could be beneficial. Clear guidance (such as by law or professional guidelines) for various healthcare providers offering genomic information will allow proportional resource commitment and optimized, efficient care, communication and support for patients and their families.

Additionally, further integrating other healthcare professionals and supporting staff, such as genetic counsellors and psychologists, was identified as a crucial strategy to better support patients as they navigate the complex communication process. Genetic counsellors and psychologists can play a crucial role as mediators between patients and clinical geneticists. One participant reported how stigma and practical burdens of pursuing additional care may deter some patients from utilizing counselling and psychological support. They suggested therefore that these professionals should be integrated in the consultation sessions as early as possible to reduce the likelihood of this occurring. Additional resources are also needed to help educate healthcare professionals outside of the genetics specialty in how to address family communication.

E-Health solutions emerged as a potential avenue for further exploration. Proposed strategies included the creation of materials to guide patients on how to inform at-risk family members and foster conversations. More specifically, the creation of tools to assist patients in communicating with their family members, such as role play videos and a set of versatile 'tell the family tools' (with several modules for different patients, family members, genetic conditions, and contexts), were suggested as a means to cater to diverse patient needs. These could be made available to patients through an online platform. Ensuring equitable access to these tools would be necessary, particularly in this context for individuals who may face challenges in accessing digital modules (Phillips et al., 2020).

Conclusion

Given the familial implications of genetic testing and genomic sequencing data, it is imperative to strike a balance between the rights and responsibilities of patients, at-risk relatives, and healthcare professionals. Based on our workshop discussions with interdisciplinary and international experts, we present several challenges stemming from the intricate process of family communication of genetic risk information, which will become more acute as the mainstreaming of genomics advances. Developing adequate guidance—in the form of not just guidelines and policies, but also through education and engagement—for healthcare professionals and patients is crucial to better navigate this process. Enhancing pre-test counselling and follow up procedures, implementing policies to clarify roles and responsibilities, and training healthcare professionals (both within and outside genetics services) are essential steps to address concerns related to communicating genetic risk in families.

References

Andersson A, Hawranek C, Öfverholm A, et al. Public support for healthcare-mediated disclosure of hereditary cancer risk information: Results from a population-based survey in Sweden. Hered Cancer Clin Pract. 2020;18(1):1-13. doi:10.1186/S13053-020-00151-0/FIGURES/5

Black L, McClellan KA, Avard D, et al. Intrafamilial disclosure of risk for hereditary breast and ovarian cancer: points to consider. J Community Genet. 2013;4(2):203-214. doi:10.1007/s12687-012-0132-y

Brownsword R & Wale J. The right to know and the right not to know revisited: Part One. Asian Bioeth Rev. 2017;9(1-2):3-18. doi:10.1007/S41649-017-0012-1/METRICS

Clarke A, Richards M, Kerzin-Storrar L, et al. Genetic professionals' reports of nondisclosure of genetic risk information within families. Eur J Hum Genet. 2005;13(5):556-562. doi:10.1038/sj.ejhg.5201394

Daly MB, Montgomery S, Bingler R, et al. Communicating genetic test results within the family: Is it lost in translation? A survey of relatives in the randomized six-step study. Fam Cancer. 2016 Oct;15(4):697-706. doi: 10.1007/s10689-016-9889-1.

D'audiffret Van Haecke D & de Montgolfier S. Genetic Test Results and Disclosure to Family Members: Qualitative Interviews of Healthcare Professionals' Perceptions of Ethical and Professional Issues in France. J Genet Couns. 2016;25(3):483-494. doi:10.1007/S10897-015-9896-7

D'audiffret Van Haecke D & de Montgolfier S. Genetic diseases and information to relatives: practical and ethical issues for professionals after introduction of a legal framework in France. Eur J Hum Genet. 2018;26(6):786-795. doi:10.1038/s41431-018-0103-9

Derbez B. Is there a "right time" for bad news? Kairos in familial communication on hereditary breast and ovarian cancer risk. Soc Sci Med. 2018 Apr;202:13-19. doi: 10.1016/j.socscimed.2018.02.022. Epub 2018 Feb 23. PMID: 29500986.

Dheensa S, Fenwick A, Lucassen A. Approaching confidentiality at a familial level in genomic medicine: a focus group study with healthcare professionals. BMJ Open. 2017;7(2):e012443. doi:10.1136/bmjopen-2016-012443

Dheensa S, Lucassen A, Fenwick A. Limitations and Pitfalls of Using Family Letters to Communicate Genetic Risk: A Qualitative Study with Patients and Healthcare Professionals. J Genet Couns. 2018;27(3):689-701. doi:10.1007/s10897-017-0164-x

Dove ES, Kelly SE, Lucivero F, et al. Beyond individualism: Is there a place for relational autonomy in clinical practice and research? Clin Ethics. 2017;12(3):150-165. doi:10.1177/1477750917704156

Gaff CL, Clarke AJ, Atkinson P, et al. Process and outcome in communication of genetic information within families: a systematic review. Eur J Hum Genet. 2007;15(10):999-1011. doi:10.1038/sj.ejhg.5201883

Geelen E, Van Hoyweghen I, Horstman K. Making genetics not so important: family work in dealing with familial hypertrophic cardiomyopathy. Soc Sci Med. 2011;72(11):1752-1759. doi:10.1016/j.socscimed.2010.06.012

Gilbar R & Barnoy S. Facing legal barriers regarding disclosure of genetic information to relatives. New Genet Soc. 2020;39(4):483-501. doi:10.1080/14636778.2020.1755639

Hawranek C, Hajdarevic S, Rosén A. A Focus Group Study of Perceptions of Genetic Risk Disclosure in Members of the Public in Sweden: "I'll Phone the Five Closest Ones, but What Happens to the Other Ten?" J Pers Med 2021, Vol 11, Page 1191. 2021;11(11):1191. doi:10.3390/JPM11111191

Li S-T, Sun S, Lie D, et al. Factors influencing the decision to share cancer genetic results among family members: An in-depth interview study of women in an Asian setting. Psycho Oncology. 2018;27(3):998-1004. doi:10.1002/pon.4627

Lucassen A. Should families own genetic information? Yes. BMJ. 2007;335(7609):22. doi:10.1136/bmj.39252.386030.AD

Lucassen A & Gilbar R. Alerting relatives about heritable risks: the limits of confidentiality. 2018;361:k1409. doi:10.1136/bmj.k1409

Meggiolaro N, Barlow-Stewart K, Dunlop K, et al. "Disclosure to genetic relatives without consent – Australian genetic professionals' awareness of the health privacy law." BMC Medical Ethics 2020

Mendes Á, Paneque M, Clarke A, et al. Choosing not to know: accounts of non-engagement with pre-symptomatic testing for Machado-Joseph disease. Eur J Hum Genet. 2019;27(3):353-359. doi:10.1038/s41431-018-0308-y

Menko FH, Aalfs CM, Henneman L, et al. Informing family members of individuals with Lynch syndrome: a guideline for clinical geneticists. Fam Cancer. 2013 Jun;12(2):319-24. doi: 10.1007/s10689-013-9636-9.

Middleton A, Patch C, Roberts J, et al. Professional duties are now considered legal duties of care within genomic medicine. Eur J Hum Genet. 2020;28(10):1301-1304. doi:10.1038/s41431-020-0663-3

Otlowski M.F. Australian reforms enabling disclosure of genetic information to genetic relatives by health practitioners. J Law Med. 2013;21(1):217-234.

Otlowski, M.F. Disclosing genetic information to at-risk relatives: new Australian privacy principles, but uniformity still elusive. Medical Journal of Australia, 2015 202: 335-337. https://doi.org/10.5694/mja14.00670

Parker M & Lucassen AM. Genetic information: a joint account? BMJ. 2004;329(7458):165-167. doi:10.1136/bmj.329.7458.165

Pedrazzani C, Aceti M, Schweighoffer R, et al. The Communication Chain of Genetic Risk: Analyses of Narrative Data Exploring Proband–Provider and Proband–Family Communication in Hereditary Breast and Ovarian Cancer. J Pers Med. 2022;12(8):1249. doi:10.3390/JPM12081249

Petersen HV, Frederiksen BL, Lautrup CK, et al. Unsolicited information letters to increase awareness of Lynch syndrome and familial colorectal cancer: reactions and attitudes. Fam Cancer. 2019;18(1):43-51. doi:10.1007/s10689-018-0083-5

Phillips, A., Vears, D.F., Van Hoyweghen, I., et al. (2020). Digital tools for sharing genetic information with family members. Lancet Oncology, 21 (7), 891-892. doi: 10.1016/S1470-2045(20)30106-6

Phillips A, Borry P, Van Hoyweghen I, et al. Disclosure of genetic information to family members: a systematic review of normative documents. Genet Med. 2021;23(11):2038-2046. doi:10.1038/s41436-021-01248-0

Phillips, A., Dewitte, I., Debruyne, B., et al. (2023). Disclosure of genetic risk in the family: A survey of the Flemish general population. EJMG, 66 (8), Art.No. ARTN 104800. doi: 10.1016/j.ejmg.2023.104800

Phillips, A., Bronselaer, T., Borry, P., et al. (2022). Informing relatives of their genetic risk: an examination of the Belgian legal context. European Journal Of Human Genetics. doi: 10.1038/s41431-021-01016-3

Rothstein MA. Reconsidering the duty to warn genetically at-risk relatives. Genet Med. 2018;20(3):285-290. doi:10.1038/gim.2017.257

Suthers GK, Armstrong J, McCormack J, et al. Letting the family know: Balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder. J Med Genet. 2006;43(8):665-670. doi:10.1136/jmg.2005.039172

van den Heuvel LM, Smets EMA, van Tintelen JP, et al. How to inform relatives at risk of hereditary diseases? A mixed-methods systematic review on patient attitudes. J Genet Couns. 2019;28(5):1042-1058. doi:10.1002/jgc4.1143

Young AL, Butow PN, Tucker KM, et al. Challenges and strategies proposed by genetic health professionals to assist with family communication. Eur J Hum Genet. November 2019. doi:10.1038/s41431-019-0447-9

Annex

The interdisciplinary expert panel was composed of the following participants:

- Dr. Carla van El, Post-doctoral Researcher, Amsterdam UMC (The Netherlands)
- Prof. Dr. Helle Vendel Petersen, Hvidovre Hospital (Denmark)
- Dr. Christine Patch, Genetic Counselor, Wellcome Genome Campus (UK)
- Dr. Danya Vears, Post-Doctoral Researcher, Murdoch Children's Research Institute (Australia)
- Dr. Colin Mitchell, Head of Humanities, PHG Foundation (UK)
- Prof. Dr. Emmanuelle Rial-Sebbag, Professor in Health Law, University of Toulouse (France)
- Prof. Dr. Ainsley Newson, Professor in Bioethics, University of Sydney (Australia)
- Prof. Dr. Sandrine de Montgolfier, Professor in Bioethics Université Paris-Est Créteil (France)
- Dr. Alvaro Mendes, Post-doctoral researcher, University of Porto (Portugal)
- Dr. Laurent Pasquier, Clinical Geneticist, CHU Rennes (France)
- Prof. Dr. Anna Rosén, Clinical Geneticist, Umea University (Sweden)
- Prof. Dr. Eline Dancet, Professor in Health Communication, KU Leuven (Belgium)
- Prof. Dr. Stefaan Callens, Professor in Health Law, KU Leuven (Belgium)
- Prof. Dr. Hilde Van Esch, Clinical Geneticist, UZ/KU Leuven (Belgium)
- Dr. Saskia Bulk, Clinical Geneticist, CHU Liège (Belgium)
- Dr. Wannes Van Hoof, Sciensano (Belgium)
- Dr. Ann Gils, Director Prevention and early detection, Kom op tegen kanker (Belgium)
- Prof. Dr. Pascal Borry, Professor in Bioethics, KU Leuven (Belgium)
- Prof. Dr. Ine Van Hoyweghen, Professor in Medical Sociology, Leuven Centre for Health Humanities, KU Leuven (Belgium)
- Dr. Amicia Phillips; Post-Doctoral Researcher, KU Leuven/University of Exeter (Belgium/UK)
- Dr. Janneke Kuiper, Postdoctoral Fellow, Leuven Centre for Health Humanities, KU Leuven (Belgium)
- Maria Siermann, PhD candidate, Centre for Biomedical Ethics and Law, KU Leuven (Belgium)
- Dr. Eva Van Steijvoort, Post-Doctoral Researcher, Centre for Biomedical Ethics and Law, KU Leuven (Belgium)