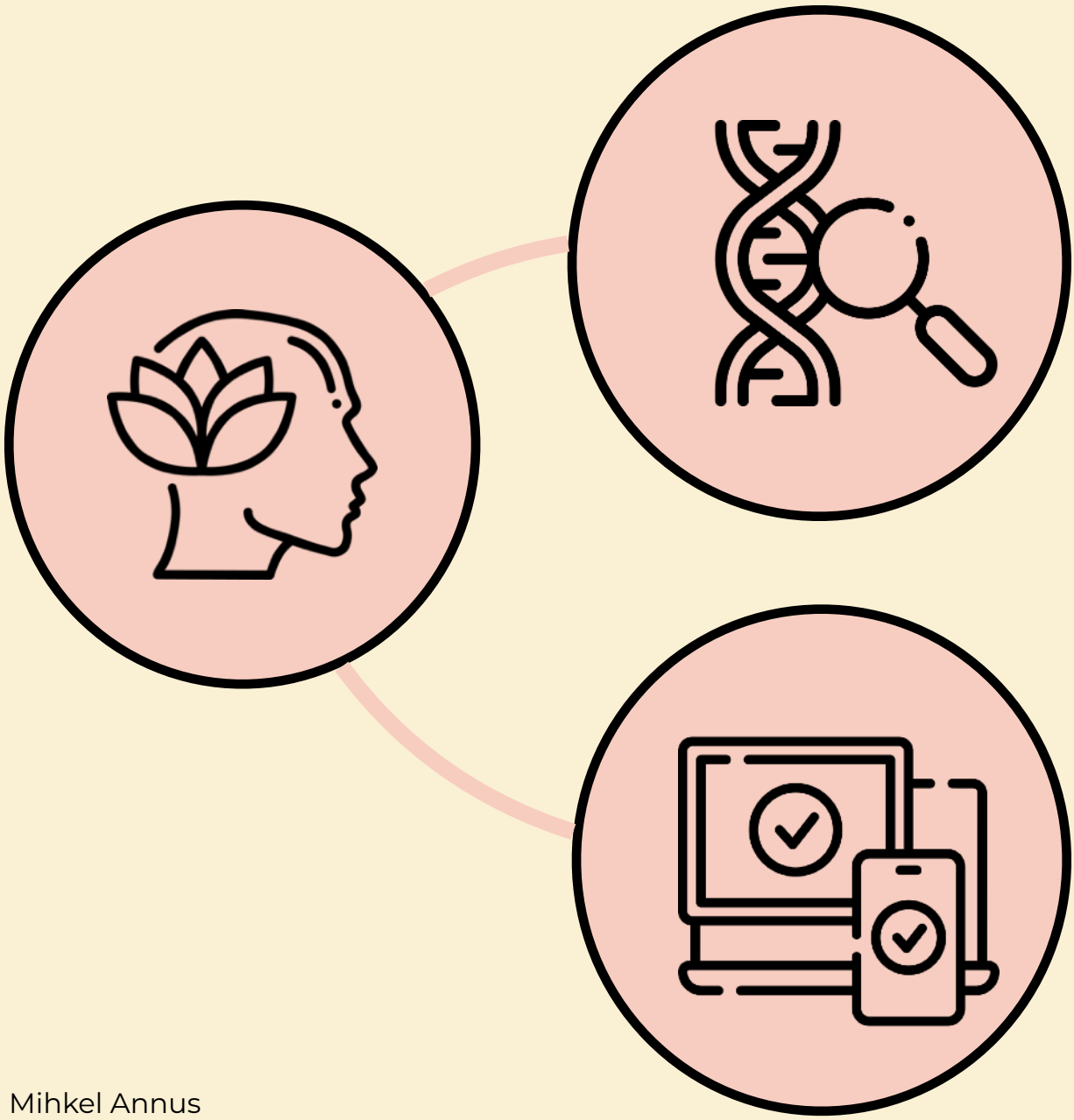
MASTER'S THESIS

Digital communication of polygenic risk for breast cancer to female recipients

A service design approach



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MSc Strategic Product Design

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Master's thesis

Digital communication of polygenic risk for breast cancer to female recipients:
A service design approach

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Disclaimer: The Master's thesis is not affiliated with the Estonian Health Insurance Fund or the Estonian Biobank.

Acknowledgements

Dear reader,

Thank you for taking the time to have a look at my Master's thesis about digital communication of polygenic risk for breast cancer to female recipients. To date, this has been the biggest research and design project I have worked on, so I am excited to share it with you.

Here, I would like to take the opportunity to thank my supervisors Lianne and Marijke for their rigorous and extensive contributions in guiding me along this journey. You supported but also challenged me like great supervisors should. You have enabled me to significantly push beyond the research and design skills I had before starting this graduation project. Furthermore, you were attentive and flexible in helping me organize my graduation project amidst time limitations.

I want to express gratitude to my family for supporting me during this rewarding but also demanding process. Throughout the thesis, you were always there to listen and help. You helped me to sort my thoughts and focus on the important. You consistently motivated me to take a break every once in a while, get some fresh air, and exercise. And I think I made a difference, for both my mental and physical health.

A further thank you to contact persons from the Estonian Health Insurance Fund and the Estonian Biobank, who helped me to become aware of the topic and who gave me valuable insights on designing for the healthcare system in Estonia. You helped me better understand how to handle research ethics in a sensitive domain, which will be very valuable for me in future work.

I also want to thank my friends and peers for being there for me. After a few days of concentrated writing at home, it was liberating to catch up and enjoy the company.

This graduation project concludes my time as a Master's student. Thank you for making this journey possible.

Mihkel Annus
Delft, 2024

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Executive summary

Technological advancements in genomic analysis have enabled the use of polygenic risk scores (PRS) to determine an individual's genetic predisposition to various diseases. The Estonian Health Insurance Fund (EHIF) is planning to introduce a service where 40-year-old women in Estonia can receive an indicator of their polygenic risk for breast cancer (BC). If a high PRS is detected, the individual is enrolled in the breast cancer screening program to help identify potential cancer earlier.

Although PRS itself does not pose a direct health threat, its communication may cause worry and lead recipients to seek support from healthcare professionals, adding strain to the healthcare system. Therefore, EHIF aims to convey PRS results through a digital channel that helps recipients understand their results without causing undue worry or prompting unnecessary consultations.

This graduation project focuses on the digital communication of BC PRS, aiming to provide recipients with peace of mind regarding their genetic risk. A literature review was conducted to analyze genetic risk, existing PRS services, and the informational and social support needs of recipients. To further understand the needs of 40-year-old women in Estonia, the target group for this service, 15 user and expert interviews were conducted.

Based on the insights from the interview studies and the literature reviews, the following aspects from genetic counseling are highlighted to create peace of mind for recipients receiving a BC PRS result:

- Use of empathic statements,
- reflective interaction with the recipients and clarification of PRS information,
- use of analogies,
- providing a sense of control to the recipient,
- framing genetic risk as a small change, and
- highlighting the value of PRS communication as an early warning.

As part of the conceptualization phase, a service blueprint and two roadmaps were developed. The service blueprint proposes the My Genetic Mirror concept, aiming to create an identity-focused perception of the service, simplify the PRS result using a fire safety metaphor, spread out information delivery to prevent overwhelming communication and provide resources for follow-up questions.

To ensure the future development of the service concept, both a strategic and a tactical roadmap were formulated. These roadmaps envision creating emotionally reassuring yet cost-efficient genetic health management, fostering peace of mind.

A preliminary evaluation of the service concept was conducted with four design students. The thesis concludes by highlighting contributions to new knowledge, identifying limitations, providing recommendations, and offering a personal reflection.

1. Introduction

This chapter introduces the project background and context, highlights its goals, the relation to the faculty of Industrial Design Engineering, and highlights the main stages of the project.

1.1 Background

1.2 Project context

1.3 Goals of the project

1.4 Relation to the faculty of Industrial Design Engineering

1.5 Project overview

1.1 Background

Advancements in the field of genomic testing and analysis have resulted in methods to provide individuals with insights about their genome. Since the human body's susceptibility to disease is at least partly determined by the genome, genomic testing has the potential to give an early warning about diseases that may develop in the future (Dudbridge, 2013). At the same time, however, healthcare costs are rising. This seeks healthcare providers to deliver services in more efficient ways, including digital channels.

The Estonian Health Insurance Fund (EHIF) is proposing the launch of a personalized screening program targeting women at the age of 40. A polygenic risk score (PRS) is a numerical estimate of an individual's genetic predisposition to a disease, calculated from the combined effects of multiple genes (Lewis & Vassos, 2020). Women with a high polygenic risk score are invited to begin mammographic screening at 41, while for others the standard BC screening program is offered from age 50, consistent with current practice. (estPerMed I, 2021) A constraint of the program, however, is that the information is intended to be delivered digitally. Therefore, when the information is delivered, no medical professional will be immediately available in person, thus it is crucial that the recipients clearly understand their situation to make informed decisions and reduce potential confusion and anxiety. Thus, EHIF's main goal for the program is to have more accessibility in communicating genetic risk, since providing in-person genomic counseling for every participant is limited in feasibility and viability. Furthermore, EHIF aims to limit the influx of women, and thus further strain, on the healthcare system.

This thesis explores the development and implementation of a digital communication tool designed to convey polygenic risk scores (PRS) for breast cancer (BC) to women. Given that BC is the leading cause of death among women aged 20-50 globally (Iacoviello et al., 2021), understanding and effectively communicating genetic risk is crucial. Personalized healthcare (also known as precision medicine) refers to a medical model that tailors healthcare decisions and treatments to the individual characteristics of each patient. This approach considers the patient's genetic makeup, lifestyle, and other factors to provide more precise, predictive, and effective healthcare (Goetz & Schork, 2018). The integration of PRS into personalized healthcare can enhance early detection and preventive strategies, potentially improving outcomes for high-risk individuals. (Roberts et al., 2023; Mavaddat et al., 2019)

Despite the potential benefits, the communication of PRS poses significant challenges. Current genetic counseling involves face-to-face interactions where medical professionals provide explanations and emotional support. However, the digital format potentially lacks this immediate personal touch, raising concerns about recipients' understanding and emotional reactions to their genetic risk information. This thesis aims to address these challenges by developing a digital tool that not only effectively communicates PRS but also provides the

necessary informational and emotional support by leveraging communication strategies used by genetic counselors. The overarching aim of the thesis is to support the process of making genetic risk information available to wider audiences.

1.2 Project context

This project was undertaken with the goal of being proposed to the Estonian Health Insurance Fund (EHIF), which is currently leading the development of a personalized breast cancer screening that includes providing a recipient with a test result for polygenic breast cancer risk. In case of high polygenic risk, the individual would be invited to the Estonian nationwide breast cancer screening program from age 41 instead of the current 50. (TAI, n.d.; estPerMed I, 2021).

EHIF's initiative relates to developments from the gene donation program of the Estonian Biobank (estPerMed I, 2021). The Biobank, a part of the Institute of Genomics at the University of Tartu, is a population-based biobank that collects genetic and health data from Estonian residents. Established in 2000, the Biobank aims to improve public health through research and personalized medicine. It currently houses genetic data from over 200,000 individuals, which constitutes about 20% of the Estonian adult population (Leitsalu et al., 2015a). The gene donation program of the Estonian Biobank invited volunteers to donate blood samples, from which DNA is extracted and analyzed. The collected data is anonymized and used for research to identify genetic risk factors for common diseases, improve the understanding of genetic influences on health, and develop personalized treatment and prevention strategies (Leitsalu et al., 2015b). The collaboration between EHIF and the Estonian Biobank aims to incorporate these genetic insights into public health initiatives, including the personalized breast cancer screening program. (estPerMed I, 2021) The project brief is included in Appendix E. For purposes of clarity, no collaboration agreement between the TU Delft and EHIF or the Estonian Biobank was signed for this research project.

1.3 Goals of the project

The goal of this project is to create a service blueprint and a respective strategic roadmap for communicating genetic risk via a digital service. These two deliverables can be used by healthcare providers, such as EHIF, as inputs for implementing digital polygenic risk communication tools. In the context of academic research, the project aims to inform personalized healthcare initiatives, such as PROPHET (Pastorino et al., 2024) and BRIGHT (BRIGHT - EIT Health, 2022) of the European Union in further developing the theoretical framework of genetic counseling and polygenic risk communication.

1.4 Relation to the faculty of Industrial Design Engineering

This thesis contributes to academic literature on the intersection between design and genetic counseling. It focuses on the goal "Design For Our Future" of Industrial Design Engineering (IDE) by exploring how strategic and interaction design can help to communicate genetic risk to a wider audience. In turn, the findings of this thesis can provide help for healthcare systems in the future to potentially detect disease, including breast cancer, earlier.

1.5 Project overview

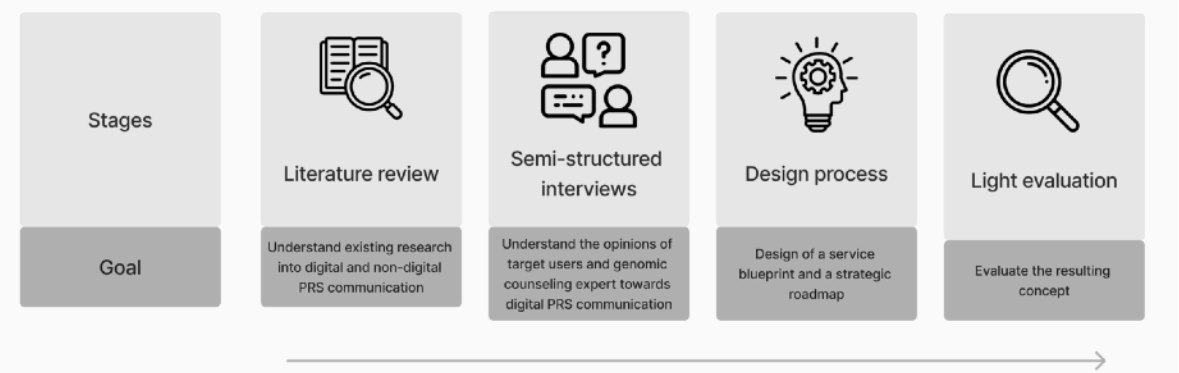


Figure 1: Visualization of the project stages and the respective goals.

The project was organized into four main stages: literature review, qualitative semi-structured interviews, design process, and a light evaluation procedure. A literature review was conducted to understand previous research into genetic risk communication, including polygenic risk. The goal was to understand which informational and social support recipients of PRS need to understand their health situation. The qualitative semi-structured interviews were done with the aim of understanding the target group's attitude towards the proposed digital PRS communication service and their related informational and social support needs. The interview insights served as input for determining the focus points of the subsequent design process. The interviews were performed with both members of the target group of the proposed service as well as academic experts in genetic counseling. The target group consists of 40-year-old women in Estonia receiving a digital PRS result for BC.
The design process followed the double-diamond process (Kochanowska & Gagliardi, 2022). First, this included initial diverging and subsequent converging in the problem space aimed at specifying a relevant problem statement. Secondly, another phase of diverging and converging in the solution space was performed to choose the solution deemed most desirable, feasible, and viable. To gather feedback and perform an iteration on the design

solution, a light evaluation was organized with four participants. Figure 1 shows a visualization of the project stages.

2. Literature review

This chapter introduces the findings from the conducted literature review into breast cancer, genetic risk, its communication, and respective user needs. The chapter concludes by highlighting the identified gap in the literature.

2.1 Breast cancer

2.2 Genetic breast cancer risk

2.3 Polygenic risk scores in the Estonian healthcare system

2.4 Existing services

2.5 Current polygenic risk score communication

2.6 Summary of the literature review and gap

2.1 Breast cancer

Breast cancer (BC) is a metastatic cancer that can transfer to other, distant organs, including the bone, liver, lung, and brain. The spreading is the main factor that causes BC incurability. (DeSantis et al., 2016) In 2020, BC caused over 685,000 deaths globally (World Health Organization, 2023).

Most BC appears in women, as there are about 100 BC cases in women for every one case in men (Siegel et al., 2017). BC is the primary reason for death among women aged 20-50 years worldwide (Iacoviello et al., 2021).

There are various risk factors for BC. These include gender, age, estrogen levels, previous family history, genetic risk factors, and an unhealthy lifestyle (Majeed et al., 2014). According to the WHO (2023), about half of all BCs appear in women who do not have particular risk factors beyond their gender and age. Egen et al. (2017) highlight that lower socioeconomic status is consistently linked to increased cancer mortality across all racial and ethnic groups. It is noted that an early BC diagnosis can result in a favorable prognosis and a high rate of survival. In North America, timely detection of the disease is paired with a five-year relative survival rate of over 80%. (DeSantis et al., 2016)

2.2 Genetic breast cancer risk

There are two common ways of measuring an individual's genetic risk for BC: monogenic risk and polygenic risk scores. Monogenic risk in breast cancer refers to the increased risk of developing breast cancer due to mutations in a single gene. These mutations are usually inherited and can significantly elevate an individual's risk compared to the general population (Turnbull & Hodgson, 2005).

On the other hand, PRS considers mutations in multiple genes. PRS are combinations of genetic variations that capture an individual's susceptibility to diseases. (Lewis & Vassos, 2020). PRS are calculated by combining the weighted sum of associated alleles within each subject. For this, a set of single nucleotide polymorphisms (SNP) are used. Each SNP represents a variation in a single DNA building block, which is called a nucleotide. When the function of these alleles is combined, it is statistically significantly associated with BC incidence. (Dudbridge, 2013; Wang et al., 2018) However, it must be noted that PRS does not capture the entire genetic risk of an individual (Lewis & Vassos, 2020).

The extent to which PRS accounts for the genetic predisposition to BC ranges from approximately 5 to 40% (Singh et al., 2016; Wittersheim et al., 2015; van Marcke et al., 2016; Aloraifi et al., 2015) with a typical range between 15-20% (estPerMed I, 2021). By integrating PRS into risk evaluation models, a more tailored risk assessment can be achieved. In the context of BC, PRS can help to identify women with a higher likelihood of developing the

disease, and in turn, facilitate the adoption of stratified screening and prevention approaches. (Roberts et al., 2023; Mavaddat et al., 2019) A systematic review by Dixon et al. (2022) suggests that cancer screening informed by polygenic risk is likely to be more cost-effective compared to other options, referring to more conventional screening methods based on broader population-level criteria such as gender, age, and family history.

2.3 Polygenic risk scores in the Estonian healthcare system

The mammographic BC screening program organized by the public Estonian Health Insurance Fund (EHIF) covers women aged 50-69 and 74 years; However, studies show that some women outside this age group have a significantly increased risk of morbidity (Partridge et al., 2016; Korde et al., 2015). While managing monogenic mutations linked to moderate and high risk of hereditary BC has become a standard practice clinically, PRS testing has not been done before in Estonia. (estPerMed I, 2021)

This begs the question, how should the participants of the personalized BC screening program make sense of their health situation, and be provided the necessary informational and social support through a digital tool? The aim of the following section was to investigate existing research in supporting women in receiving a personalized BC risk score.

2.4 Existing services

Company	**Key focus**	**Design insights**
Antegenes	Comprehensive process, personalized strategies	Personalized prevention plans for providing assistance to recipients, ensure that a non-medical person can understand the results
23andMe	Direct-to-consumer, broad accessibility	Use of different visualization methods
Myriad Genetics	Extensive testing, detailed risk assessment	Include detailed, actionable reports
Invitae	Multi-gene panels, integration into clinical practice	Ensure that the PRS communication is useful for both the recipient and that it can also be used by a medical professional in providing support

Table 1: Overview of the findings from existing solutions in digital PRS communication.

Examples of services from different countries that communicate PRS to counselees are in the private sector (Table 1). As of the author's knowledge, there is no similar solution introduced in the public sector. This section aims to explore and critically analyze private-sector examples of PRS communication services to extract insights for designing a similar solution in the public sector.

Figure 2.: Genetic PRS test report cover page from NalaGenetics (Aldila et al., 2023).

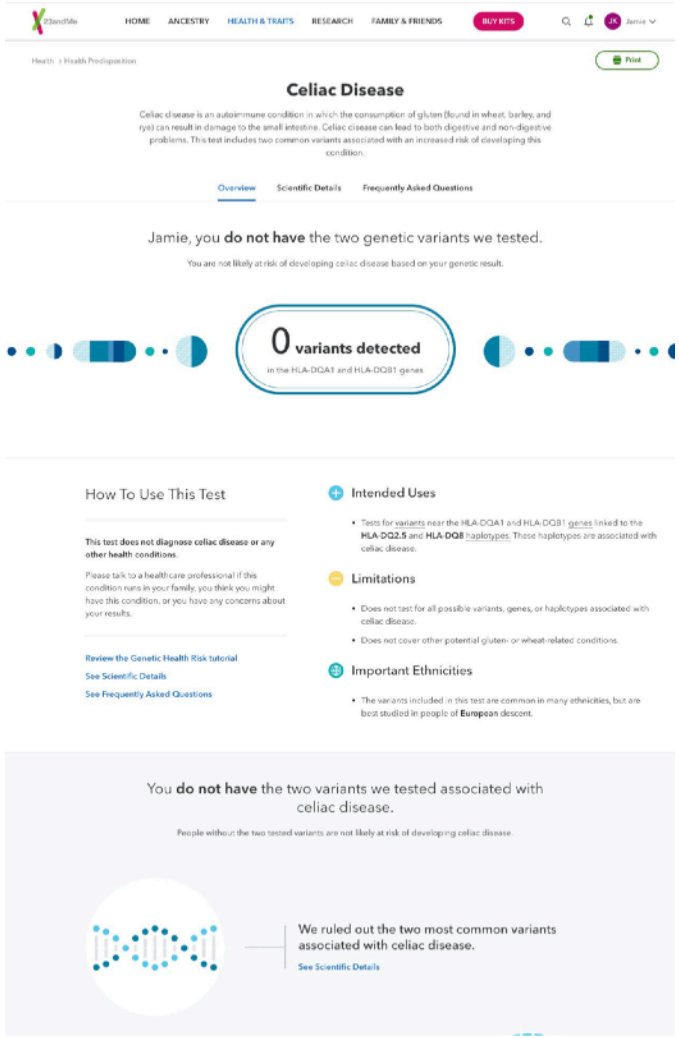


Figure 3: Example genetic test report for celiac disease by 23andMe (Sample 23andMe Report for Genetic Health, n.d.).

Figure 4.: Genetic PRS test report (written in Estonian) cover page from the Estonian company Antegenes (Antegenes, 2024).

Antegenes

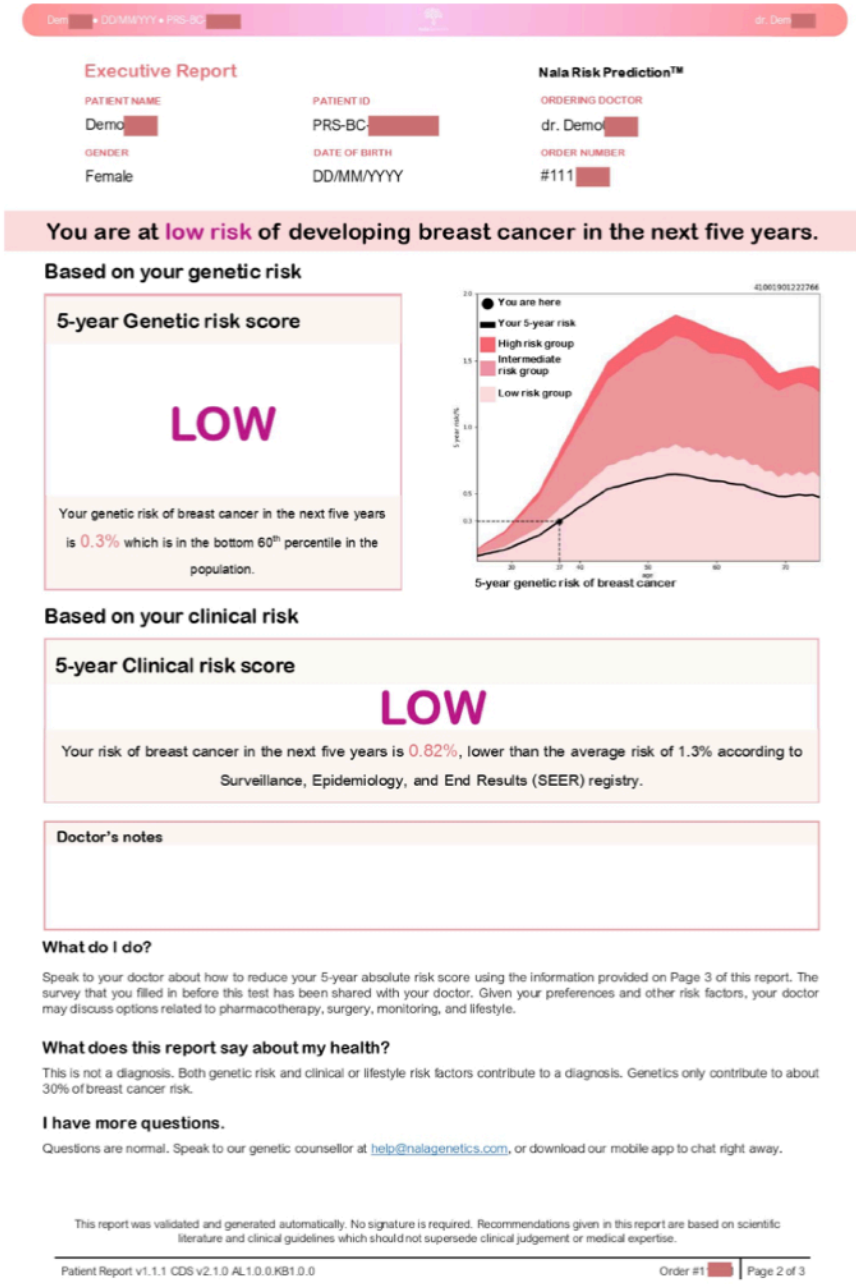
One notable provider is the Estonian company Antegenes, which offers a comprehensive BC precision prevention service. This service targets women aged 30 to 75 and includes a detailed process where a genetic test is conducted, analyzed, and communicated to the patient. The test involves the analysis of 2803 genetic variations associated with increased susceptibility to breast cancer. The results are used to create personalized prevention strategies, which can include lifestyle changes and monitoring. Antegenes' approach emphasizes the integration of genetic data with clinical guidance to improve patient outcomes and empower individuals with actionable health insights. Although Antegenes' PRS report at times summarizes the information in an easy-to-understand manner, it still retains a focus on accuracy and includes many instances of medical jargon. (Antegenes, n.d.) This information may be hard to comprehend for a person with a non-medical background, suggesting that the report should be discussed with a medical professional. Thus, another key learning from Antegenes' report is that the information delivery should be tailored to a non-medical person.

23andMe

In the United States, 23andMe offers a direct-to-consumer genetic testing service that includes a report on breast cancer risk. 23andMe analyzes several genetic markers associated with BRCA1 and BRCA2 genes, among others, to assess an individual's risk. Their user interface uses a variety of visuals and the report is designed to be read by a person with a non-medical background, thus providing a user-friendly value proposition with accessibility for a variety of numeracy levels. The report provides customers with insights into their genetic predispositions, which they can discuss with their healthcare providers for further action. While 23andMe focuses on making genetic information broadly accessible, it recommends that PRS recipients consult a healthcare professional on their own to help with interpreting the result, requiring additional effort by the user (23andMe, n.d.).

Myriad Genetics

Myriad Genetics, another US-based company, offers more comprehensive testing through its MyRisk Hereditary Cancer test. This test evaluates multiple genes, identifying variations that could increase the risk of breast cancer and other hereditary cancers. Myriad Genetics provides detailed risk assessment reports (written with medical jargon) that include recommendations for managing and mitigating risk, which are designed to be used by healthcare providers in developing personalized care plans for patients. Their service is particularly valuable in clinical settings where a thorough understanding of genetic risk can inform treatment and prevention strategies (Myriad Genetics, n.d.). However, the provider is



Executive Report

Nala Risk Prediction™

PATIENT NAME: Demo [redacted]
PATIENT ID: PRS-BC [redacted]
ORDERING DOCTOR: dr. Demo [redacted]
GENDER: Female
DATE OF BIRTH: DD/MM/YYYY
ORDER NUMBER: #111 [redacted]

You are at low risk of developing breast cancer in the next five years.

Based on your genetic risk

5-year Genetic risk score

LOW

Your genetic risk of breast cancer in the next five years is 0.3% which is in the bottom 60th percentile in the population.

Based on your clinical risk

5-year Clinical risk score

LOW

Your risk of breast cancer in the next five years is 0.82%, lower than the average risk of 1.3% according to Surveillance, Epidemiology, and End Results (SEER) registry.

Doctor's notes

What do I do?

Speak to your doctor about how to reduce your 5-year absolute risk score using the information provided on Page 3 of this report. The survey that you filled in before this test has been shared with your doctor. Given your preferences and other risk factors, your doctor may discuss options related to pharmacotherapy, surgery, monitoring, and lifestyle.

What does this report say about my health?

This is not a diagnosis. Both genetic risk and clinical or lifestyle risk factors contribute to a diagnosis. Genetics only contribute to about 30% of breast cancer risk.

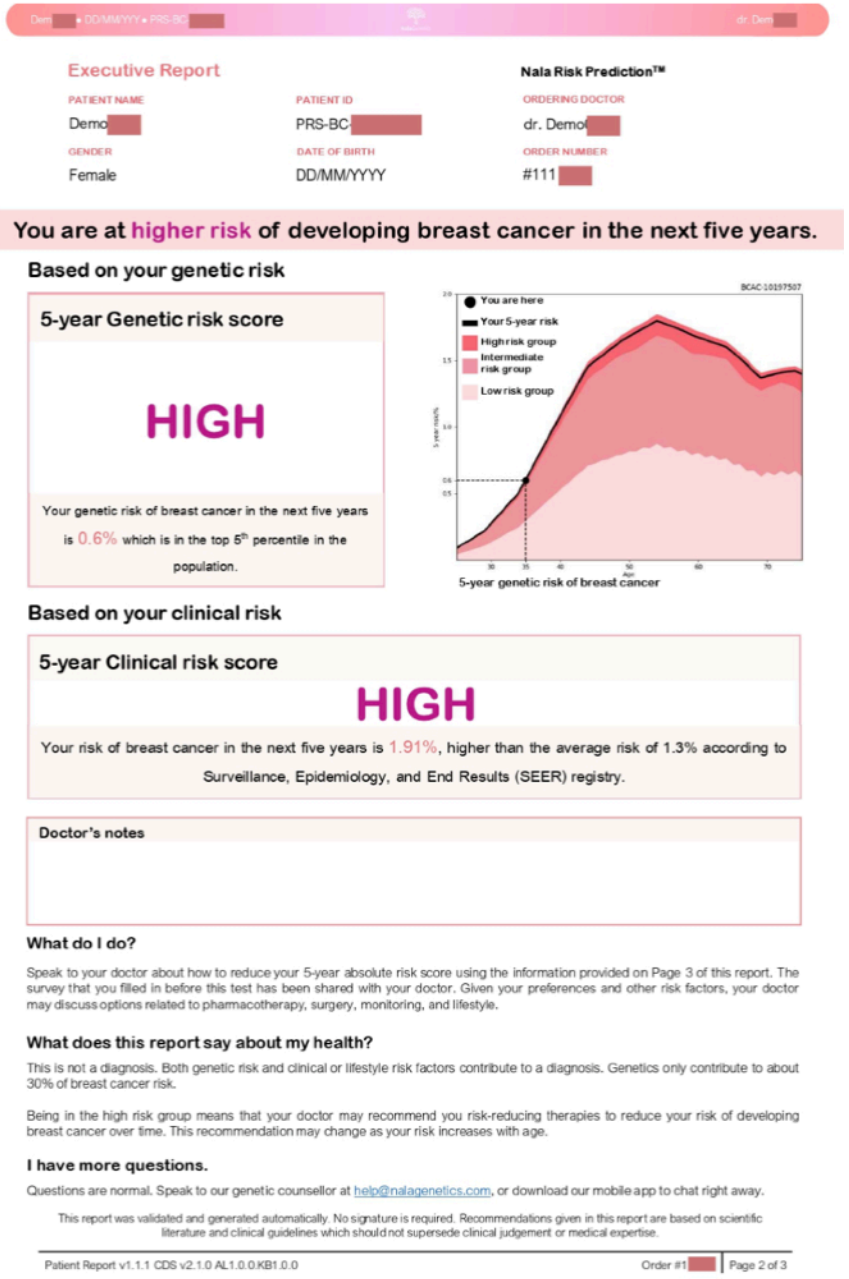
I have more questions.

Questions are normal. Speak to our genetic counsellor at help@nalagenetics.com, or download our mobile app to chat right away.

This report was validated and generated automatically. No signature is required. Recommendations given in this report are based on scientific literature and clinical guidelines which should not supersede clinical judgement or medical expertise.

Patient Report v1.1.1 CDS v2.1.0 AL1.0.0.KB1.0.0 | Order #1 [redacted] | Page 2 of 3

Figure 5: Second page of the low PRS result report from NalaGenetics (Aldila et al., 2023).



Executive Report

Nala Risk Prediction™

PATIENT NAME: Demo [redacted]
PATIENT ID: PRS-BC [redacted]
ORDERING DOCTOR: dr. Demo [redacted]
GENDER: Female
DATE OF BIRTH: DD/MM/YYYY
ORDER NUMBER: #111 [redacted]

You are at higher risk of developing breast cancer in the next five years.

Based on your genetic risk

5-year Genetic risk score

HIGH

Your genetic risk of breast cancer in the next five years is 0.6% which is in the top 5th percentile in the population.

Based on your clinical risk

5-year Clinical risk score

HIGH

Your risk of breast cancer in the next five years is 1.91%, higher than the average risk of 1.3% according to Surveillance, Epidemiology, and End Results (SEER) registry.

Doctor's notes

What do I do?

Speak to your doctor about how to reduce your 5-year absolute risk score using the information provided on Page 3 of this report. The survey that you filled in before this test has been shared with your doctor. Given your preferences and other risk factors, your doctor may discuss options related to pharmacotherapy, surgery, monitoring, and lifestyle.

What does this report say about my health?

This is not a diagnosis. Both genetic risk and clinical or lifestyle risk factors contribute to a diagnosis. Genetics only contribute to about 30% of breast cancer risk.

Being in the high risk group means that your doctor may recommend you risk-reducing therapies to reduce your risk of developing breast cancer over time. This recommendation may change as your risk increases with age.

I have more questions.

Questions are normal. Speak to our genetic counsellor at help@nalagenetics.com, or download our mobile app to chat right away.

This report was validated and generated automatically. No signature is required. Recommendations given in this report are based on scientific literature and clinical guidelines which should not supersede clinical judgement or medical expertise.

Patient Report v1.1.1 CDS v2.1.0 AL1.0.0.KB1.0.0 | Order #1 [redacted] | Page 2 of 3

Figure 6: Second page of the high PRS result report from NalaGenetics (Aldila et al., 2023).

reliant on in-person interactions with a healthcare professional. This suggests that their PRS information is not tailored for the recipient to understand on their own.

Invitae

Invitae provides genetic testing services that focus on breast cancer risk through multi-gene panels. These panels test for mutations in a broad range of genes associated with breast cancer, providing a comprehensive genetic risk profile. Invitae emphasizes the integration of genetic information into clinical practice, offering support for healthcare providers to interpret the results and guide patients in making informed decisions about their health. Their approach combines advanced genetic testing with clinical expertise to enhance patient care and preventive strategies (Invitae, n.d.). Like Myriad Genetics, Invitae (US) is also reliant on reports with medical jargon and on interactions with healthcare professionals to help understand the PRS result.

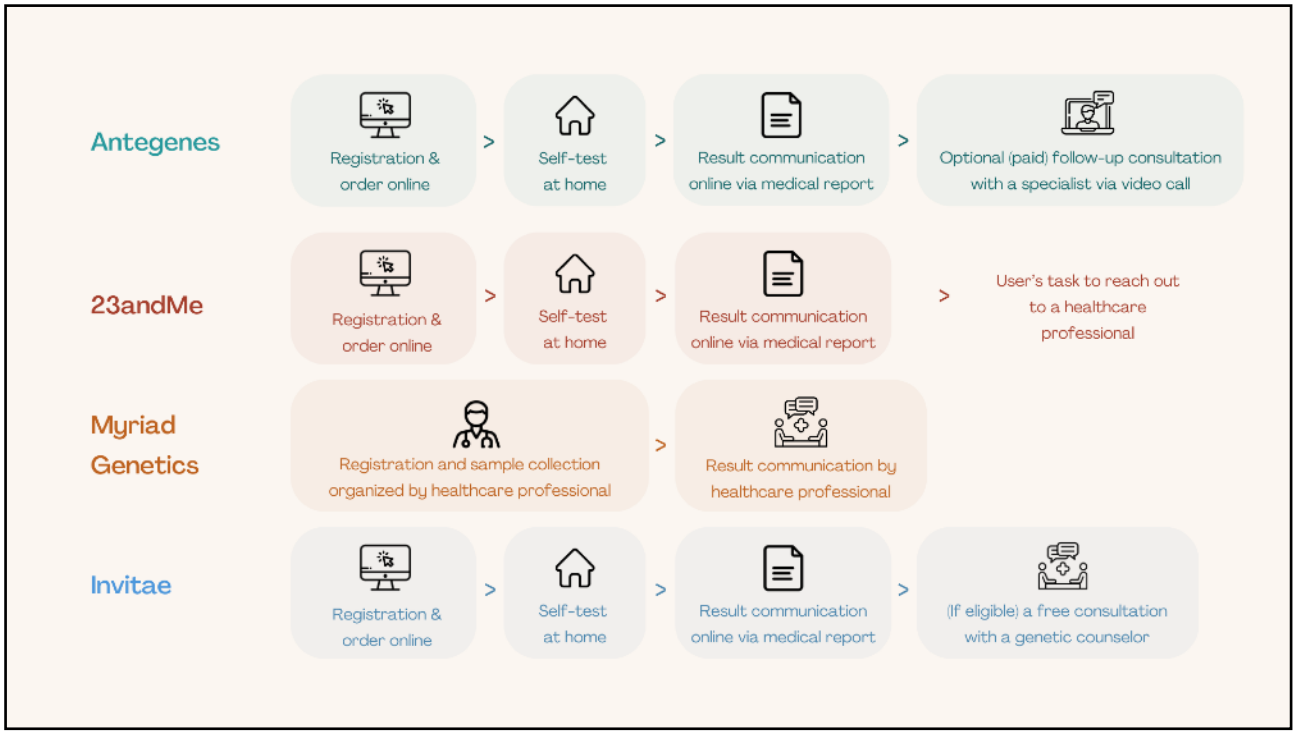


Figure 4b: Overview of the service modules of Antegenes, 23andMe, Myriad Genetics, and Invitae.

Differences in service modules

The service modules of the four companies have overlapping parts with differing aspects (Figure 4b). For customers of Antegenes, 23andMe, and Invitae, participation in the service starts via an online online and registration. The customer is sent a self-test for a saliva sample, which is then sent back to the company for analysis. Subsequently, the result is communicated online with a medical report. Antegenes and Invitae customers can choose to do a follow-up consultation to discuss the result. For 23andMe customers, no consultation is included,

meaning that the recipients need to take up contact with a healthcare professional on their own to discuss the result.
Compared to the other companies, Myriad Genetics organizes their service through a healthcare professional, who takes care of registering the user to the service and the sample collection process. The PRS is communicated in a consultation with a healthcare professional. Thus, a consultation is included in the service. This makes Myriad Genetics much more reliant on healthcare professionals for organizing the service and communicating PRS results.

Considerations

Across all companies, the emphasis is on personalized prevention strategies and integrating genetic data with clinical guidance. The design solution should ensure that PRS results are communicated in a way that takes into account the person's health profile, with clear guidance on lifestyle changes, monitoring, and preventive measures. Services like 23andMe highlight the importance of accessibility and user-friendly interfaces, thus suggesting that the design should prioritize ease of use and accessibility. Companies like Myriad Genetics and Invitae demonstrate the value of comprehensive genetic testing and detailed risk assessments, indicating that the solution should provide detailed reports with actionable recommendations. However, all four companies rely on medical reports and healthcare professionals for communicating genetic risk. This may suggest that the information used by healthcare professionals is not tailored for PRS recipients with additional interpretation. Thus, it is important to consider how the digital communication channel could communicate PRS information in a way that is understandable (i.e., that does not rely on medical jargon) to the recipient, and providing support that does not give a reason for the PRS recipients to consult a healthcare professional to understand their result.

2.5 Current polygenic risk score communication

The communication of a polygenic risk score is typically done in the presence of a medical professional (when offered by companies in the private sector), who assists the woman in understanding the report, answering follow-up questions, and providing social support (e.g., Aldila et al., 2023, and Leitsalu et al., 2022). A goal of genetic counseling is to inform women about the different types of risk factors that contribute to their health (Bredart et al., 2021). Gupta et al. (2021) highlight that during the consultation the focus is mainly on informational needs.
Example PRS test reports by Aldila et al. (2023) (Figure 2), 23andMe (23andMe, n.d.) (Figure 3), and Antegenes (Figure 4; in Estonian) include the following main parts: explanation of how a person should understand the risk, how the result was determined, what the risk level is, how

it compares to the wider population, suggestions what the counselee should do to manage their risk, and contact details in case of questions. Figures 5 and 6 (Aldila et al., 2023) highlight the respective low and high-risk reports.

2.5.1 Informational support needs

To understand the informational support that literature highlights as valuable to PRS recipients, multiple articles on the topic were explored.

Informational support need	Explanation
Personal information	The information should be tailored to the individual receiving it (Gorman et al., 2022; Brockman et al., 2021; Amornsiripanitch et al., 2017)
Understandable information	The information should be made understandable regardless of a person's educational background and previous experience with the healthcare system (Gorman et al., 2022; Aldila et al., 2023; Bredart et al., 2021; Evans et al., 2015).
Meaningful information	The information should provide helpful value to the individual (Yanes et al., 2020; Mason et al., 2023).
Trustworthiness	The individual should trust the results regarding data security, accuracy, ethnic diversity-related aspects, and evidence-based guidelines (Mason et al., 2023; Lewis et al., 2022).
Risk self-management	The individual should receive follow-up steps (action plan) after receiving the PRS result (Gorman et al., 2022; Aldila et al., 2023; Huang & Perri, 2014).

Table 2: A summary of informational support needs for PRS delivery highlighted in the literature.

The literature highlights multiple informational needs of PRS counselees, highlighted in Table 2. Most importantly, BC risk information should be personal, understandable, meaningful (Gorman et al., 2022), trustworthy, and relate to self-management. In the following parts, the different informational support needs are elaborated.

Personal information

A key finding by Gorman et al. (2022) is that BC information should be personal. This entails that the delivered information should be tailored to the recipient's specific characteristics, such as their genetic background, lifestyle, and behaviors, to increase perceived relevance and understanding. Moreover, personalization was found to relate to reduced anxiety, increased confidence in understanding BC information, and willingness to consider appropriate risk management pathways.

Brockman et al. (2021) highlight that counselees had different levels of informational needs and, in turn, wanted information at different depths. For instance, a participant noted that it would be desirable if there were a choice for the amount of information received. Amornsiripanitch et al. (2017) highlight that, for instance, the higher the patient's risk, the more information they want on their situation. Furthermore, the counselees tend to receive more information if they are accompanied by a family member in an in-person consultation (MacDonald et al., 2007).

Understandable information

Another finding by Gorman et al. (2022) entails that the provided information has to be understandable. Aldila et al. (2023) suggest that more comprehensive results are desirable. A preference to receive more detailed information is proposed by Gorman et al. (2022). They highlight that women prefer knowing in detail about how each specific risk factor contributes to the overall risk.

Conversely, information overload and medical jargon should be avoided, especially with counselees with lower education levels (Bredart et al., 2021; Evans et al., 2015; Aldila et al., 2023).

Suggested interventions include visual communication tools (Forrest et al., 2019; Brockman et al., 2021), paired written and numerical information with graphical presentation (Hamilton et al., 2020), avoidance of overly medical information, and including simpler language in the report, e.g., on grade 8 literacy level (Evans et al., 2015; Farmer et al., 2020; Kaur et al., 2018).

Meaningful information

Meaningful information relates to providing the counselees with helpful value. Yanes et al. (2020) suggest that women prefer to receive PRS more if they perceive greater benefits and fewer barriers. Mason et al. (2023) inquired the question "Does knowing genetic risk actually help the patient?". Although some respondents mentioned that they would want to know PRS information, others felt that it would not be helpful unless a certain definitive action could be taken to make healthy choices. It was further highlighted that learning about one's PRS may not significantly influence one's current lifestyle or health-related decisions.

Trustworthiness

Mason et al. (2023) highlight participants' uncertainty about PRS risk score trustworthiness. These concerns relate to three aspects: data security, accuracy, and diversity. Data security concerns entail risks relating to genomic data storage and potential misuse. Accuracy relates to the notion that the results should have very high accuracy to be considered useful in the first

place. Diversity concerns highlight how a lack of genomic diversity in the development of PRS methodologies can result in increased inequalities in health outcomes, especially for ethnic minorities.
Lewis et al. (2022) highlight the role of evidence-based guidelines in risk management. They conclude that evidence-based guidance is important for getting counselees to change their health-related habits. According to the study, some counselees would only be willing to act on PRS information when evidence-based guidelines are present.

Risk self-management

Counselees want to know whether risk factors are modifiable. However, few people are aware of the link between lifestyle and cancer risk. (Gorman et al., 2022) To manage one's BC risk, follow-up steps are desired (Aldila et al., 2023).
In taking action to manage one's risk, multiple factors influence the counselee's behavior. A study about decision-making after a genetic test (in the case of Alzheimer's; Huang & Perri, 2014) found that prediction value, availability of treatments, and anonymity and confidentiality play a role. First, prediction value highlights how confident counselees can be about the test's accuracy. Secondly, the availability of treatments refers to interventions that would help counteract the risk of disease. Thirdly, anonymity and confidentiality were highlighted, which entails concerns about discrimination in employment and health insurance.

2.5.2 Social support needs

Next to informational support needs, the literature also highlights social support needs for receiving a PRS result. Social support needs relate to the emotional and instrumental support in the context of social interactions of a BC PRS counselee with the aim of improved mental health (Finfgeld-Connett, 2005). First, different emotional reactions to being communicated a PRS result are highlighted, followed by the social support needs identified from the literature (Table 3), and respective elaborations.

Emotional reactions to receiving PRS results

Unless specified otherwise, the following findings relate to PRS communication in an in-person consultation with a medical professional. Most of the women receiving low-risk reports mentioned that they were relieved (Aldila et al., 2023; reports based on both PRS and non-genetic risk factors). There was a consensus among participants receiving a low-risk report that engaging in more frequent consultations with medical professionals was desirable

to ensure that the risk remains low. However, it was also reported that no significant changes would occur in the relationship with their physician.

Aldila et al. (2023) highlight that almost all participants receiving high-risk reports said they would feel afraid, shocked, surprised, sad, worried, and anxious. Yanes et al. (2021) found that a higher PRS was associated with greater distress and decisional regret towards participating in the communication. Although Leitsalu et al. (2022) agree that receiving information about high PRS can be uncomfortable, these feelings were not long-lasting. Lewis et al. (2022) further suggest that patients do not experience long-term negative psychological effects after receiving unfavorable genetic results. Nonetheless, counselees receiving a high-risk result reported that their awareness of BC would be increased (Aldila et al., 2023). On the contrary to agreeing to receive PRS results, women declining participation in a PRS communication experience more decisional regret, fewer perceived benefits, and greater concerns about PRS (Yanes et al., 2021).

Van Erkelens et al. (2017) studied the emotional response of women aged 50-74 to an online self-test for BC risk determination. It was found that taking an online self-test to determine BC risk decreases anxiety and BC risk perception while distress scores remain unchanged. The participants would fill in a questionnaire before and after going through the test. The self-test would include respondents entering the family history of BC, ovarian, and prostate cancer for themselves as well as for close relatives, after which a three-tier risk level was communicated. The participants in the high-risk cohort were directly advised to consult with a genetic counselor. Participants in the medium-risk cohort were recommended annual mammography starting from 40 or 50 years and the low-risk cohort was recommended to continue in the screening program.

Social support need	**Explanation**
Tailored amount of support	Counselees require a variable amount of social support (Gupta et al., 2021; Yanes et al., 2021).
Communication strategy usage by genetic counselor	Counselees prefer the medical professional to be empathic in their statements (Gupta et al., 2021; Lobb et al., 2005; Gorman et al., 2022).
Opportunity to inquire	The counselee should be able to ask questions to clarify the results (Yanes et al., 2021; Gupta et al., 2021; Kaur et al., 2018).
Varied perspectives on the result	The counselee prefers to receive further perspectives on the PRS, in addition to a medical professional (Aldila et al., 2023; Young et al., 2017).

Table 3: A summary of the main social support needs highlighted in the literature. The references are included in the following sections.

Tailored amount of support

As with informational needs, social support needs vary by individual, e.g., as shown in consultation length and associated level of risk (Gupta et al., 2021; Yanes et al., 2021). Claes et al. (2004) describe that receiving test results can be stressful in the short term. Young et al. (2017) found that most women were relieved to receive their risk profiles, with a minority feeling anxious immediately due to confirmation of their risk of second primary cancer. Further reasons for anxiety include feelings of guilt for passing on mutations to offspring. However, receiving the PRS tends to vindicate previous risk management decisions and clarify one's risk.

In general, participants generally considered the results valuable, understandable, and not scary (Leitsalu et al., 2022). Receiving PRS is further associated with increases in confidence and a sense of personal control (Wallingford et al., 2023).

Communication strategy usage by genetic counselor

Gupta et al. (2021) outline that genetic consultations utilize different communication strategies, such as empathic statements, reflection of feeling and content (e.g., statements to give reassurance and validation), paraphrasing, summarizing, and using analogies to explain the PRS.

In a BC PRS consultation, the medical professional should be empathic in their statements (Gupta et al., 2021). More extensive use of empathic statements is associated with lower levels of stress after consultation with a genetic healthcare provider (Lobb et al., 2005). Using analogies to explain the PRS can include comparing PRS to a "lottery" or a "deck of cards" (Gupta et al., 2021). Gorman et al. (2022) note that the risk estimate should be communicated in a positive tone. Gupta et al. further highlight the role of social chit-chat and laughter during observed consultations.

Opportunity to inquire

In general, counselees need help interpreting and understanding their PRS results. Thus, such support is crucial for managing any anxiety and distress associated with BC risk information. (Yanes et al., 2021) Thus, the counselee should be invited to ask questions and share their thoughts on the received PRS and general BC risk (Gupta et al., 2021). Dijkstra et al. (2013) warn that a counselor's verbal dominance leads to lower perceived needs fulfillment. According to Yanes et al. (2021), women overestimate their level of risk for BC, which can be an additional factor to inquire about the PRS result.

The need to ask follow-up questions was also evident in a study by Kaur et al. (2018) – reading an informational leaflet about BC PRS testing at home left some participants worried and upset. These worries related to issues understanding the leaflet, fear about test results showing

higher-than-anticipated risk as well as concerns about implications of the test results for the person's descendants.

Varied perspectives on the result

Female counselees prefer to discuss the result with multiple contact persons to receive further perspectives. In addition to a medical professional, this can include family members and friends, especially if they have a medical background or have previously been diagnosed with BC (Aldila et al., 2023). Counselees consider PRS results affecting their family (Young et al., 2017).

2.6 Summary of the literature review and gap

In summary, informational and social support needs are critical for supporting BC PRS counselees. Informational support is typically delivered through a report, which includes the risk level, an explanation of the risk level, how the risk score is likely to develop throughout life, which strategies there are for self-management, and which support pathways are available. Social support is provided to a woman by the medical professional, but also by family, friends, and coworkers. However, the medical professional has the crucial role of consulting and comforting the patient during the consultation.

Current research has mostly been done under the assumption that a medical professional is in the room to assist the counselee. However, healthcare systems, including in Europe, are under increasing pressure due to demographic change, chronic diseases, and declining budgets (Marschang et al., 2015). Thus, to avoid overwhelming the healthcare system and increasing wait times, a digital solution for a younger target group (40-year-old women) in a public sector context is proposed. Existing studies by van Erkelens et al. (2017) and Keane et al. (2021) suggest that digitally communicating BC PRS risk scores can yield positive psychological outcomes for participants, including reduced anxiety and lower BC risk perception. However, studies have highlighted the value of face-to-face consultations (Leitsalu et al., 2022), and differing levels of support needs, especially in cases of higher risk (Gupta et al., 2021; Yanes et al., 2021).

Thus, the following is unknown:

- to which extent BC PRS communication can be performed digitally for women younger than the current BC screening program starting age of 50,
- how to develop a digital tool that women understand, trust, and can use independently without needing to see a medical specialist (and thus avoid strain on the healthcare system),

- and how to ensure the digital communication tool provides adequate social support to women, helping them feel supported and reassured, even without immediate access to a medical specialist.

3. Interview studies into support needs for polygenic risk score communication

This chapter highlights the interview studies conducted with potential users and experts in BC PRS communication. For this, the aim of the interview studies, the outline, and the method are elaborated. The results from both the user and expert studies are formulated and discussed.

3.1 Aim of the interview study and outline

3.2 Method

3.3 Results from the user interview study

3.4 Results from the expert interview study

3.5 Discussion

3.6 Overall conclusion of the user and expert studies

3.1 Aim of the interview study and outline

The aim of the interview study was to understand the informational and social support needs of the target group of the service. The target group comprises 40-year-old women receiving a BC PRS in the context of a digital service. The study sought to identify what specific types of information and social support these women require when receiving their PRS, in order to inform the design of a digital, human-centered service concept that effectively addresses these needs.

The interview study was based on this research question:
What informational and social support do women receiving a polygenic risk score for BC need in the context of a digital communication tool?

The interview study part of the thesis included a semi-structured qualitative interview study. In total, 15 interviews were conducted (Table 4). There were two groups of interview participants recruited: 35-43-year-old women in Estonia with no previous diagnosis of breast cancer (potential users) and Estonian genomics experts with experience in communicating BC PRS (the experts). The potential users group included at least three individuals who had voluntarily participated in the gene donation program of the Estonian Biobank. All interviews were conducted in Estonian. All interviews were audio recorded, with additional written notes by the researcher.
Ethical approval was received for the study by the Human Research Ethics Committee of the Delft University of Technology (Approval #3942). Bound by the consent forms gathered during the interviews, access to the interview data is restricted to the researcher and the supervisory team, with the interview data intended for erasure three years after the study's completion.

3.2 Method

3.2.1 Participants in the user study

In the user study group, 12 interviews were conducted (Table 4). Of those, 10 were in-person, with two online due to better fit in scheduling. Estonian women were chosen as the sample since they constitute the current target group of the eventual design intervention. The recruitment followed a referral sampling (network sampling) strategy where the participants were contacted via suggestions from the researcher's own personal network. Qualitative

interviews were chosen as they are more suited for rich information and for discussing sensitive topics with no judgment (Dempsey et al., 2016).

Procedure

The interviews ranged from 25 to 75 minutes, with most remaining between 30 and 45 minutes, which was the planned duration of the interviews. In cases where an interview went overtime, the researcher asked the participant whether they were willing to extend the interview time.

During the interviews, the researcher first asked the participants general healthcare-related questions, e.g., what they think of the digital Health Portal in Estonia and who they usually contact in case they get sick. Next, the participants were asked to imagine that they received an invitation to take part in a personalized breast cancer screening program by the Estonian Health Insurance Fund. The researcher explained the concept of the service, what the procedure looks like, and what potential results can be. Then, the participants were asked to express what reactions they would expect to have in case they were to receive a low and respectively, a high BC PRS result. The mock-up reports were given to the participants separately, one after the other. In the last part, the participants were given a mock-up report of both a low and a high-risk PRS result and were asked to again share their expected reactions in case they would receive such a score in real life. The mock-up reports were based on the high and low-risk reports by Aldila et al. (2023), translated into Estonian. The translation process was aided by a general medical specialist.

If the mock-up result was perceived as inducing discomfort for the interview participant, the researcher suggested sharing how another person would react to getting such a score. With two interview participants, the high and low-risk BC PRS reports were not discussed since the interview had gone over the expected interview duration.

3.2.2 Participants in the expert study

The participants in the expert study (Table 4) included respondents from Estonia with academic and professional backgrounds in the fields of biomedicine and genetic research. Their expertise encompassed communicating BC PRS through consultations. Three interviews with experts were conducted, of which two were online (due to better schedule suitability) and one in-person. The expert sample consisted of two women and one man. The professional backgrounds of the participants include biomedicine and genetic technology, a researcher with a background in providing genetic feedback to recipients, and a professor with a relation to medical genetics. One expert has a Master's degree, with the other

two being postdocs. These experts were affiliated with both public and private institutions involved in genomic research and medical genetics.

Interviewee	Characteristics	Knowledge about genetic risk among users
1	Office worker	Has prior experience with genetic risk analysis, understands that genetic risk scores are not diagnoses. Mentioned participating in genetic counseling and related blood tests.
2	Pharmacist, Marketing specialist, mother	Knowledgeable about different types of cancer screenings, including genetic testing. Understands the role of genetic information in assessing cancer risk.
3	Sales representative, mother	Limited knowledge about breast cancer genetics.
4	Climate advocate at an NGO, mother	Limited knowledge about breast cancer genetics.
5	Accountant, mother	Has a basic understanding of genetic risks related to breast cancer but lacks detailed knowledge. Interested in genetic screening programs.
6 (expert)	Researcher in the area of genetic feedback	n/a
7	University student, mother	Limited understanding of genetic aspects of breast cancer screening. Confused about the differences between genetic risk assessment and traditional screening methods.
8	Climate advocate at an NGO, mother	Confused about the PRS service, mistakenly associates it with mammography. Limited understanding of genetic screening.
9	Head of quality control at a small company, mother	Has heard of genetic testing and knows it involves assessing genetic risk for diseases.
10	Unemployed, married	Limited understanding of genetic risk.
11 (expert)	Researcher in the area of genetic feedback	n/a
12	Theatre producer, mother	Limited understanding of genetic risk.
13	IT development team leader, mother	Has a basic understanding of genetic risks related to breast cancer but lacks detailed knowledge.
14 (expert)	Professor, medical genetics (male)	n/a
15	Director of a public school, mother	Limited understanding of genetic risk.

Table 4: Anonymized characteristics of the participants in the user and the expert sample.

Procedure

The participants were asked about their professional background and experience in communicating BC PRS risk scores. They were asked to explain how they typically explain a BC PRS score in person, how the recipients have reacted to such consultations, and how the experts have assisted the recipient, both in providing information and offering social and emotional support.

Next, they were provided an overview of the service concept and were inquired about their feedback, critique, and suggestions on digital PRS communication. Finally, as with the main

sample, two experts were provided mock-up reports of both high and low PRS risk and were asked to provide feedback. The third expert was not inquired about the mock-up reports since the interview had gone beyond the agreed duration of the interview. The expert interview length ranged from 40 minutes to 75 minutes, with an expected duration of 30–45 minutes. If there was a high likelihood that the interview would go overtime, the participants were asked whether they were willing to extend the originally agreed-upon duration.

3.2.3 Interview data analysis

All audio recordings were transcribed verbatim and additionally replayed and checked by the researcher after the original transcribing process. Participants' identities were anonymized by giving them pseudonyms based on the sequence number of their interview, e.g., "Interviewee 1", "Interviewee 2", etc. Expert interview names are appended with "expert". After each interview was transcribed, the respective audio recording was destroyed.

The transcripts were translated into English in two steps, first using the DeepL translation tool and then the researcher manually comparing the English translation with the Estonian original and subsequently correcting the translation.

The interview data analysis was guided by the thematic analysis by Braun et al. (2023; Figure 7). First, the transcripts were coded, with similar codes then grouped using the visualization tool FigJam. Each code was put on a virtual Post-it note, with codes from the same interview being given the same color. After an extensive clustering process, the emerged clusters served as collections of similar insights. Since over 600 codes were created, it was important to perform multiple iterations in simplifying the clustering structure to formulate them as insightful sub-themes and themes. Eventually, relevant sub-themes were clustered into broader themes. The resulting code trees (for both the user and the expert studies) are highlighted in Appendices A and B.

Figure 7: An overview of the generated clusters during thematic analysis.

3.3 Results of the user interview study

This chapter serves to highlight the extracted insights relating to the research question: "What informational and social support do women receiving a personalized risk score for BC need in the context of a digital communication tool?". The results were sorted to two different parts: the general context factors to understand to help understand the beliefs and behaviors of the participants in the user study (chapter 3.3.1), followed by the identified themes (chapter 3.3.2).

3.3.1 Context factors

To understand the broader context of the relationship between the interview participants and the healthcare system in Estonia, this chapter serves as a respective overview. This includes who the participants contact in case of health problems, their relation with medical professionals, the Health Portal (in Estonian "Terviseportaal"), screening programs, and genetic testing.

First contact point in case of health problem

During the analysis, it emerged clearly that if the respondents face a health problem, their first contact point is the GP or the GP's assistant. The respondents prefer to take up contact via email, or by phone call in case of urgent matters. Some respondents are used to asking questions via the E-GP platform, which is a text-based interaction platform between a patient and a GP. If, for some reason, it is not possible to get in contact with a GP, a specialist may be approached. Furthermore, the role of family and friends as social support is also important for some respondents. They highlight that in light of a health problem, they would first discuss the situation with family and friends. However, for the vast majority of respondents, the GP's office is the first point of contact.

Relation with the GP & healthcare system in Estonia

The respondents consider the quality of help by GPs high in Estonia. Multiple respondents highlighted that in urgent situations, GPs can be reachable even on the weekend via email. As a general remark about the healthcare system, the respondents highlighted that the wait times for a doctor are long. Regarding health monitoring, regular check-ups are considered important by multiple respondents. They highlight that they are used to being in close contact with doctors about managing their health and that they consider it important.

Health Portal and test results

Respondents generally trust the e-state solutions offered in Estonia. The Health Portal is considered a significant digital touchpoint with the healthcare system. However, the respondents use the Health Portal infrequently, mainly for checking test results and setting doctor's appointments for both herself and her children. They also note problems with the Health Portal, namely difficulty in understanding medical information and jargon. Some respondents expressed frustration that they have not always received a notification about certain test results they have been expecting for a while.

Awareness and opinion about screening programs in Estonia

As the BC screening program starts from age 50, no women in the sample had experience with the program. However, multiple participants had experience with the cervical cancer (CC) screening program, which starts at age 30. They trace their awareness about the CC screening to receiving an email invitation, talking to their women's doctor, friends, seeing advertisements on the street, in sports halls and on the internet, and on the Health Portal. Awareness about BC and the BC screening program is limited. A few respondents mentioned that their main contact point with BC screening has been in noticing a mobile mammography bus on the town square and asking the staff when they are expected to take part. The topic of BC screening has come up in discussions with family and friends, who have told them about their experience participating. Some respondents also mentioned that they have been actively forwarding BC screening information to their older close relatives to encourage them to take part.

Multiple participants consider it important that parents should be role models for their children in taking part in screening programs to ensure that society takes participation in screening more seriously. It was mentioned that the older generation (older than the age group of the sample) tends to not take screening seriously enough. Millennials, the main sample of the interview study, were mentioned to have comparatively more awareness about health monitoring. However, younger generations, including Gen Z, tend to take health monitoring more seriously compared to millennials.

The respondents generally have a positive attitude towards participating in screening programs. Respondents report a feeling of certainty from having participated in a screening program and a contribution to disease prevention. Screening being covered by public health insurance was considered a further economic benefit. A respondent highlighted that although no one actively wants to go to a screening program, she considers them very necessary. Relating to the BC screening program, some respondents wish that the BC screening program would start from an earlier age due to increasing breast cancer incidence among younger women.

Motivating factors to take part in screening and the PRS-driven personalized BC screening program

Respondents highlight fear of cancer, self-efficacy and prevention, family and friends' experience, pop culture influences, and practicalities to motivate them to take part in the BC screening and the proposed PRS-driven personalized BC screening programs. Fear of cancer relates to the wish to know more about her health based on her genome. Self-efficacy and prevention relate to the feeling that she has done everything in her power to discover a potential disease as soon as possible. Family and friends' experiences relate to previous instances where a close person was diagnosed and that serves as a motivating factor for the person to take part in screening. Pop culture motivators include news about breast cancer awareness, for example, Angelina Jolie's cancer prevention therapy. Furthermore, the respondents would be motivated to take part in the PRS service due to the service being offered for free.

Participation in the Gene Donor program in Estonia

During the interviews, three respondents self-reported to have donated their genome to the Estonian Biobank as part of the Gene Donor program in Estonia, organized by the Insitute of Genomics of the University of Tartu. One respondent mentioned that she received feedback on her genome via an online video consultation and that she understood her health situation better afterward. The other two respondents said that she had donated their genomic sample a few years ago with the goal of contributing to genomic research but also to potentially get health insights about their own genome. However, they had not received any feedback after the participation. They were still expecting to receive some insights, but they were not optimistic about whether they will ever get the feedback about their genomic analysis. One of the respondents described it as "an empty experience".

3.3.2 The identified themes

In the following, the identified six themes are highlighted: Overwhelming information, overstating genetic risk, clarification about implications of the PRS result, clarification about practicalities, digital PRS communication, and mental model about PRS. Figure 8 shows an overview of the identified main themes. The code tree for the user study can be found in Appendix A.

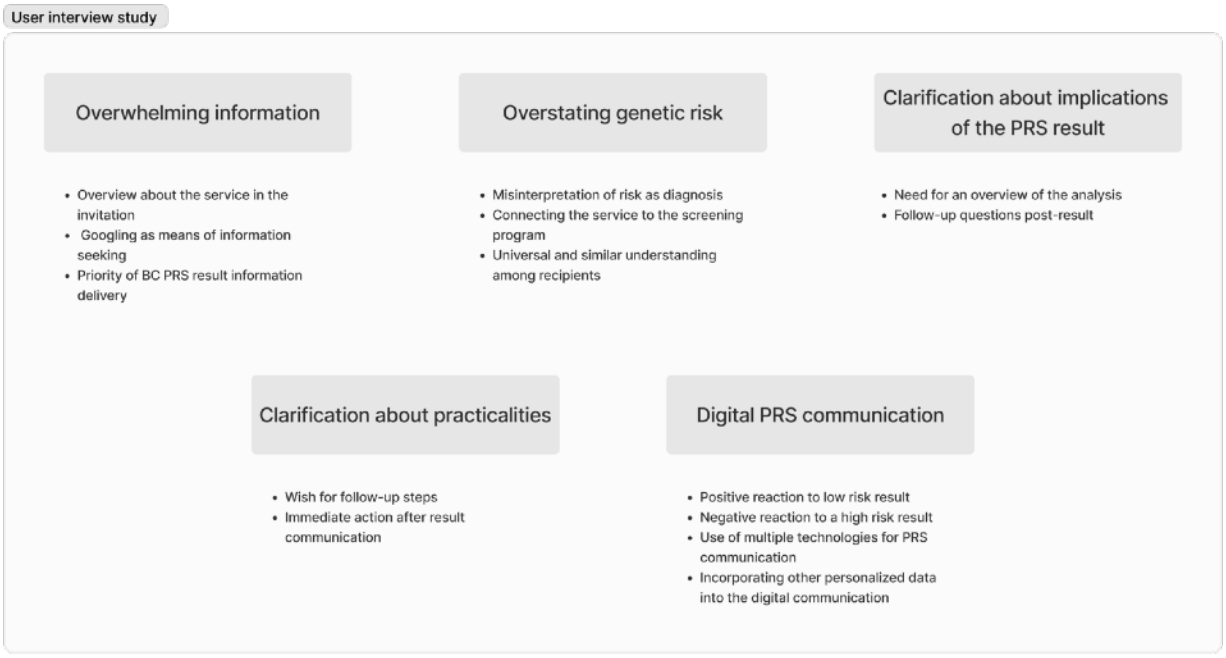


Figure 8: Overview of the themes and sub-themes identified from the user interview study.

3.3.3 Overwhelming information

The first main theme relates to PRS information likely being overwhelming for the recipient. Respondents highlight that receiving PRS information tends to induce anxiety and distress. Depending on the level of risk indicated by the PRS, the respondents may have an immediate concern about whether there is a serious health issue present. This emotional response might make them act rashly, including rushing to see a doctor to address their fears and get more clarification on their health in their own context. When reaching out to a healthcare professional, the result is perceived as urgent and with high severity. To illustrate the finding, a quote from Interviewee *5* is included:

"If a person gets like this, almost a shock, then maybe someone will go to the block [psychological shock], but I will tell you in advance that she will just start rushing, that there is something here, she tries to read, but it's like she's out of her mind and can't understand. So maybe it would be very good if it were possible to contact someone right away, that okay here is the [email] address, but... [...] that there won't be an answer right away." - Interviewee *5*

Overview about the service in the invitation

An early touchpoint between the BC PRS communication service and the recipient is in the first invitation. Since the invitation contributes to understanding about the service, the interview participants' needs and wishes were gathered for this stage. The respondents had expectations and comments about how they would prefer to be invited to the personalized BC screening program. These are highlighted in the following. The invite should be clear in terms of content, not too vague, and include information about the program. It should highlight:

- The required effort to take part in the service,
- what the procedure entails,
- how long the recipient has time to react to the invitation,
- and contact details in case of further questions,
- the goal of the study,
- why her input is necessary,
- why she is among the invitees,
- and general information materials about the service before the actual risk communication.

Upon hearing that the participant needs to provide a blood test, this aspect was deemed uncomfortable and since that they are unfamiliar with how the data will be handled throughout the genomic analysis process. Thus, the invitee would like to know what is being done with her data as part of the program.

It was also noted that if the email mentions breast cancer, it can be frightening to the recipient. Thus, it was highlighted that the invite should include information that gives a feeling of certainty. For example, the institution behind the service should be reputable for the invitee to trust the invitation.

The respondents had further questions and concerns about the name of the program and data privacy. The current draft name of the service is "Personalized breast cancer screening program". Multiple respondents were uncertain about what the word “personalized” specifically refers to in the name. Namely, how the program is personalized for every participant, which should be explained in detail. The word “program” was perceived as a long-term commitment with associated additional effort.

Googling as a means of information-seeking

After getting to know one's PRS result, some respondents highlighted the role of googling. This serves for finding follow-up information, clarification about test results, and understanding one's health conditions in general. There was one respondent who had previously received a genomic test result via the Health Portal. She said that after getting the

result via the Health Portal website, she started googling the result, which ended up confusing her. Thus, she was more eager to reach out to healthcare professionals to clarify her situation.
When going over the mock-up PRS reports during the interviews, some respondents mentioned that they would google unknown terminology in the reports. When googling about other types of medical results, the web search process was reported to be very time-consuming, misleading, and of questionable quality. Thus, some respondents avoid googling a health situation altogether. Furthermore, a person may end up googling terms before a procedure and find extreme versions of the procedure, as highlighted in the following quote.

"If plus the fact that maybe, like, when I was sent here for the examinations, the information, I actually basically googled what it all looked like and again, I googled and found the most extreme version, where it was like, what all they are going to do to me there and cut open everything, but it was actually a very standard procedure and there was nothing to be afraid of, I think that maybe the doctor could have, like, given me some information about it." - Interviewee 12

Nonetheless, some respondents consider themselves knowledgeable Google users and are used to taking the search results with skepticism.

Priority of BC PRS result information delivery

The interviews outline the preferred priority according to which respondents expect the PRS information should be presented in the result. Regarding the visual report design, large text was noticed first, with some small text deemed too small to read.
The first point of information should be the result, in a summarized format. It was mentioned that the second priority should be to communicate the probability of getting a disease and what type. Afterward, action plans such as self-management recommendations and participation in follow-up tests should be included. Furthermore, there should be additional and more specific information available in case the recipient wants to understand more about her situation.
It was mentioned that the methodology (including the analysis and the result determination processes) should remain in the last parts of the communication. A respondent mentioned that otherwise, she would get impatient when having to read through the technical methodology since that information is not relevant to her.

"I always like this version of how people start talking about [scientific] models and things [says ironically]. And then I, who have very little time, then I'm always.... Just tell me, what is it? Am I sick or not? In this sense, of course, I understand that if you want to give more in-depth information to a person and if a person wants to go deeper, some people are different from me. I'm the one who's very impatient." - Interviewee 12

3.3.4 Overstating genetic risk

The second theme relates to the finding that a PRS recipient may overstate the implications of the result. This includes misinterpreting the risk as a diagnosis and connecting the service with aspects from the BC screening program (which is a different service, e.g., including mammography screening). Moreover, it was highlighted that the information should be understood similarly for recipients with a variety of educational backgrounds.

Misinterpretation of risk as diagnosis

Multiple respondents did not differentiate between the concepts of "risk" and "diagnosis". They worried that a polygenic risk score may instead communicate a diagnosis, that the person has, or will certainly get BC in the future. This misinterpretation can lead to unnecessary anxiety and a potential rush to consult a medical professional about a health condition they do not actually have but believed might be inevitable.

“The interviewee
I rather think that a person remembers only the high risk here. She won't remember from here that this is a 5-year genetic risk score, that she will only remember something like "high".
The interviewer
That this "high" is like a bit paralyzing?
The interviewee
Yes. If we talk about the fact that it is in three groups, low, medium and high, and if I immediately see "high" from here, it can lead to the block [psychological shock] that I am now at a high risk, or do I already have [disease] in hand.
The interviewer
That you might even get the impression that it might even be a diagnosis?
The interviewee
Yes.” - Interviewee 2

Connecting the service to the screening program

When Interviewee 8 was asked to explain what she understood under "personalized breast cancer screening program", she highlighted mammography. However, mammography is part of the general breast cancer screening, but not of the PRS service at hand. Thus, it is possible that the respondent inherently mistook the PRS service for the general screening program, driven by the name including the words "screening program". This can suggest that users may not sufficiently differentiate between the PRS communication service and the BC screening program.
Mammography is a method used in general breast cancer screening programs, typically aimed at detecting early signs of breast cancer in women with no apparent symptoms. It involves X-ray imaging to identify tumors or abnormalities in breast tissue. (Gøtzsche & Jørgensen, 2013).

In contrast, a PRS service involves analyzing genetic information obtained from a blood sample. This screening method assesses an individual's genetic predisposition to developing breast cancer by examining variations in multiple genes associated with the disease. Unlike mammography, which identifies existing tumors, the PRS service aims to predict future risk based on genetic markers.
In EHIF's intended PRS communication service, the recipient first gives a blood test. The analysis of the blood test is used to determine whether the recipient is assigned for a mammography screening as part of the breast cancer screening program. The mammography is outside of the scope of the service since it does not relate to PRS communication. If a potential recipient expects mammography in the PRS communication service, it can suggest that she does not consider the PRS communication and the BC screening program as two distinct services with different methods (blood test vs mammography) and outcomes (genetic risk indication vs tumor/ abnormality identification).

"I would assume that it also includes the aspect that, a mammography has been done and nothing has been found at the moment, and also if the risk is low, then it would sound good." - Interviewee 8

Universal and similar understanding among recipients

The sample included varying educational levels. In a related way, based on the respondents' input, knowledge and understanding of the concept of genetic risk differs. Respondents highlighted on multiple occasions that medical jargon and complex elements in the mock-up PRS reports (Aldilda et al., 2023) confused them. This includes the table on the first page of both low- and high-risk reports, the meaning of "non-genetic risk", and the meaning of the "5-year risk score". Thus, the PRS result communication should be understandable for respondents from a variety of educational backgrounds.

3.3.5 Clarification about implications of the PRS result

The respondents noted that they wanted to understand the implications of the PRS result. They wish to get an overview of the results of the analysis, what it implies for one's health, and how to get answers to follow-up questions that arise in the context of the communication.

Need for an overview of the analysis results

"The first thought that would come to my mind is the question of what this high [risk] is and, what does it mean." - Interviewee 7

Respondents emphasized that the PRS communication should be simple, honest, and highlight whether there is reason for concern or not. There, the person shouldn't underestimate or overestimate her BC risk. Interviewee 12 highlighted that a recipient may not have much time to read through everything and that she wants to get a clear answer. She mentioned that she is impatient when receiving medical results and is not interested in reading about the scientific models and methods that were used to come up with the result. The respondents further highlighted the role of getting a perspective given by a human in understanding her PRS result.

The participants highlighted three informational elements that should be in the PRS communication: an overview of the analysis, non-genetic factors, and follow-up steps. First, they highlighted that a report should include an overview of what was analyzed in the genome and how one's score compares to the wider population. References for further reading were deemed important by several respondents. Next to the results from the genomic analysis, respondents aim to understand the role of non-genetic risk factors in one's overall health outlook. This involves highlighting non-genetic mitigating factors, such as lifestyle choices that can help reduce genetic risk. Upon receipt of a PRS result, some respondents had questions about what the phrasing of the risk level specifically means. There, they expect a clear explanation. If no specification is provided, they would expect to have further questions about the result. Then, further assurance about the result may be sought from a healthcare professional.

Follow-up questions post-result

Respondents highlighted a wish to access further resources in case of questions. These can include contact to a specialist or references. Some respondents mentioned that when contacting a specialist, they should have a professional specialization in genetics and thus be able to answer specific questions about PRS. In the following, different follow-up questions of recipients are highlighted in case of a low and high PRS result, respectively. Low PRS recipients would have follow-up questions about whether one should take any action and suggestions on how to keep the risk low. For further questions, contact details should be present.

High PRS recipients would highlight that a high risk would incite questions that she would prefer to have answered, in addition to receiving the report. These questions include:

1. What does the risk mean? What is wrong with my health?
2. How do I reduce my risk?
3. Are the highlighted self-management suggestions relevant to me?

Regarding question 1, recipients report confusion about what “high risk” means. Since an explanation table on the first page of the example PRS report used the terms “high risk” and “low risk” multiple times, it was confusing to recipients what the actual score was.
During high PRS communication, it should be emphasized that high risk does not constitute a diagnosis. Moreover, it was mentioned that high PRS recipients should receive as much preceding information about the results as possible, and it should be communicated quickly. This includes an explanation about what in the genome caused the high risk, how the result compares to the wider population, and how many PRS recipients with a high PRS result have actually been later diagnosed with BC.

"It's like there wasn't too much of that data, that it was just said that I'm just at a higher risk . And where did that come from? I would ask her what it was that made me have this high risk." - Interviewee 13

3.3.5 Clarification about practicalities

"I think it causes anxiety, certainly, if there's a high risk of developing breast cancer, at this given moment there's certainly only a risk, right? But still would like to have more information on what to do with it if it is a risk, how to minimize that risk as well." - Interviewee 2

Whereas the previous theme focuses on what the PRS result means, this section highlights what respondents would aim to know about the next practical steps about the result. The following sub-themes highlight the recipients' need to know about follow-up steps and that the follow-up steps should include an immediate action plan.

Wish for follow-up steps

Respondents expressed high interest in getting to know about follow-up steps after receiving the PRS result. They highlighted that there should be an action plan in place with instructions about what to do next, including lifestyle recommendations. The respondents also expressed a wish to receive a tailored screening plan. The screening plan should be based on the PRS result and the person's health profile. Interviewee 4 emphasized that the digital system should be up to date about the screening history of the person so that timely reminders could be sent.

“We find that there is a reason to invite you to the examination, for example because of your age. Or that 'too much time has passed since your last check-up', again here the question arises that the Biobank does not know when I last voluntarily went for a breast check-up. I don't know if such restrictions are very reasonable, because it means that maybe I already went to do it last month, through my gynecologist, to a private doctor.

Or at least some kind of markers that your last breast check-up was five years ago, so please come." -Interviewee 4

Respondents noted that they would also wish to see self-management recommendations about how to reduce their risk. Importantly, these recommendations should be tailored to the person, taking into account her health situation. The general self-management recommendations provided in the mock-up reports were criticized since they were not perceived as tailored to the person's health profile.

Immediate action after receiving PRS

"My first thought would be to call the GP and see if I can get a quick check-up somewhere." - Interviewee 7

Immediate action relates to a swift follow-up response that aims to clarify uncertainty about the results. An interview participant highlighted the feeling of safety – that the person knows when the next invite to a screening is coming (or alternatively the next appointment at a doctor's is scheduled) and that nothing additional needs to be done. Otherwise, the person may overreact or even panic about not getting any information and reach out to a doctor on her own (and thus further strain the healthcare system).

3.3.6 Digital PRS communication

The respondents were prompted with a potential service that would inform the person about the PRS result via a digital channel. The expected feasibility of such a service varied based on two factors: the level of risk and how well the result is explained. If the level of risk implies no further necessary action, the communication could be done digitally. However, there was concern about digital communication, since people's awareness about the topic is low and they may not know how to interpret the results. It was mentioned that the person may end up googling the meaning of the result, which can result in misinformed PRS recipients. A respondent highlighted that a person may "go crazy" before going to a doctor's if she doesn't know what the result implies.

Respondents further highlighted by that it would be desirable to familiarize oneself with the report before reaching out to a doctor. When at a doctor's appointment, forgetting the questions one wanted to ask was deemed a problem. Thus, it was mentioned that a comfortable environment, e.g., the home (and not the doctor's office), would be important for processing the information beforehand. A respondent who had previously received a BC genetic risk score and then had an online call with a doctor found the consultation useful since it clarified the situation.

Some respondents considered it important to reach out to a doctor after the PRS communication. They highlighted three advantages of face-to-face consultations for PRS

communication: human touch, more personalization, and a better reaction to emotional states. First, human touch relates to the presence of a human who assists in understanding the score, answering questions, and the person's distrust towards digital communication channels, e.g., video calls. Secondly, face-to-face consultations were seen as more personalized, with the opportunity to ask more personal questions. Thirdly, a better reaction to emotional states refers to the human ability of a doctor to tailor the consultation to suit one's psychological situation and to offer help in a supportive environment.

"Let's say that if the result is negative, that the score is very high, then yes, maybe so that it wouldn't make me too nervous or too emotional, then s/he [a doctor] should be able to manage it, I think." - Interviewee 1

The following aspects further specify the proposed insights into digital PRS communication, highlighting the differences in low and high-risk delivery, the use of different technologies for doctor-counselee interaction, as well as incorporating personal data into framing the result.

Positive reaction to a hypothetical low risk result

Respondents reacted positively to receiving a hypothetical low-risk PRS. They considered the result calming, joyful, and not give a reason to panic. Multiple respondents mentioned that in case of low risk, a doctor's consultation would not be necessary or that the doctor's role can remain minimal. A respondent brought an example from experience with a women's doctor and cervical cancer screening in Estonia, that in case a result is "fine", the person is not contacted at all. However, a respondent mentioned that she would prefer to have a consultation in case of low risk.

The expected behavior of recipients after low PRS communication relates to general self-efficacy as well as self-monitoring and screening. General self-efficacy entails the person understanding that there is no reason to worry about her health situation and reading about self-management suggestions. Further health monitoring relates to the person expecting to continue participating in screening programs and self-monitoring her body as usual. In general, multiple respondents highlighted that they would not do much about the situation.

Negative reaction to a hypothetical high risk result

Upon receiving a hypothetical high PRS result, respondents indicated a range of behaviors and reactions. Respondents reported a variety of negative emotions such as anxiety, fear, worry, shock, and paranoia after receiving a high-risk result, with the recipient possibly thinking of the worst-case scenario (e.g., cancer diagnosis) and only understanding a limited part of the PRS result.

A respondent highlighted that communicating the PRS during the workday can be dangerous, as it may hamper the person's ability to concentrate and focus, especially if she works a delicate or dangerous job. Multiple participants expressed the need for a clear solution to manage their high PRS, such as self-management strategies and detailed screening plans, with recommendations that would motivate them to change their habits and improve their health. Multiple respondents mentioned they would urgently contact a doctor for a consultation, or they anticipated that their doctor would reach out to them. It was noted by a respondent that doctors typically contact patients only if there is something abnormal in the test results. If required to visit a doctor's office to learn about their PRS result, respondents felt they would experience significant stress due to not knowing the result. Moreover, it was noted that being invited to a doctor's appointment to receive a PRS result would be very stressful since one is used to receiving medical results online. Thus, if the person had to go to a doctor, the recipient may think that the result is so "bad" that it is important that it's done in person. Some respondents wished to get advice on how frequently they should participate in screening, and there was also a desire for more comprehensive testing to better understand their health status. Participants emphasized the importance of a prompt response from the healthcare system, including quick access to a doctor's consultation, although one respondent noted that not everyone might be motivated to proactively contact a doctor. Having additional tests, either through regular screening or private clinics, was considered desirable by some respondents, and self-monitoring of health was also mentioned as a way to anticipate potential diseases.

Multiple respondents preferred an in-person consultation, needing certainty, knowing that someone cares about them, and the inclusion of human touch when a PRS is communicated. It was noted, however, that they would prefer to go over the result first online and then have the option to reach out to a doctor.

"[In response to how the follow-up steps should be communicated to the PRS recipient] Don't get in touch with your doctor, because we'll be calling for an examination here in a year's time anyway. And that you're in no hurry. But if it says everywhere here to contact your doctor, that your doctor will give you more information and your test results are forwarded to your doctor. Obviously, I want to know what my doctor thinks about this, because he is my body doctor after all.

[...] Yes, you can't say don't contact your doctor, that's not what I'm reading out here, that maybe if you're not in a hurry, you don't have to rush, you're not dying of cancer with high probability, you're just in a risk group, right, you've got time and you're already being [contacted]." - Interviewee 13

Use of multiple technologies for PRS communication

Multiple technologies for doctor-counselee interaction emerged from the interviews, including video materials, phone calls, video calls, emails, and AI agents. First, video materials were proposed as a digital medium that can help the counselee understand PRS information.

This could include videos to explain the concept of PRS as well as the meaning of the result. Secondly, phone calls were regarded as a way of communicating the first crucial information. However, phone calls are considered shorter than video calls. If a lot of information is communicated in a phone call, the recipient can also forget it more easily.
Thirdly, video calls are considered more high level than phone calls and they are more suitable for asking follow-up questions. Benefits include less traveling to the doctor's, especially if a person lives in a rural area. A respondent mentioned that it would be a comfortable way of contacting the doctor next to raising her child at home. Many respondents are used to or imagine themselves video-calling a doctor, although some prefer in-person consultations.
Fourth, emails were considered useful for remembering an invitation to screening since they leave a concrete record. However, contact via email was deemed limited and some respondents mentioned that email invites are easier to forget. Fifth, AI agents like ChatGPT were reported to be used, including for general medical questions. Two concerns about AI were mentioned: an AI agent cannot react to humans like an actual human would, e.g., in guessing a person's non-verbal reactions, and AI agents should be trained on credible data to ensure that suggestions by the AI agent can be trusted.

[In using AI for healthcare inquiries] "The fact is that the same AI will basically lead you to the answers if the material is taken from a proper place, not some random internet thing. That it still has to be some kind of actually sorted out sort of medical information, not some kind of complete hogwash" - Interviewee 12

Incorporating other personal data into the digital communication

Respondents expected that the PRS communication would take into account their personal information like age, whether she has had children, her medical history, family history, and how long ago was the last health check-up.
The respondents may feel the need for a more tailored interpretation of their results, based on their individual health profiles (including whether she has had children, her medical history, family history, and how long ago was her last health checkup). The mock-up reports used in the user interviews may not always address individual circumstances, such as a woman's personal and family medical history, which can lead to further questions and the need for a tailored risk assessment and management plan from a doctor.
Furthermore, a differentiation of PRS recipients based on, e.g., personality type was discussed. Yet, there were ethical concerns since people part of outlier groups may be excluded:

"But in general I believe that if we have a personalised service, it has to be based on the person. Yes, I understand that we can't take it as if the archetype [interviewee's name] and the [other name] are so that we can't take it as if we can't create a kind of differentiation for each person, although that could be [good] in this

case.
But it just doesn't make sense. And then we're left with basically grouping people in this way, and I feel for people in advance who clearly don't belong in groups, because I know that one of them is definitely me, for example. I don't belong to the usual groups, and so I probably have more of these experiences than some of the others." -Interviewee 10

3.4 Results from the expert interview study

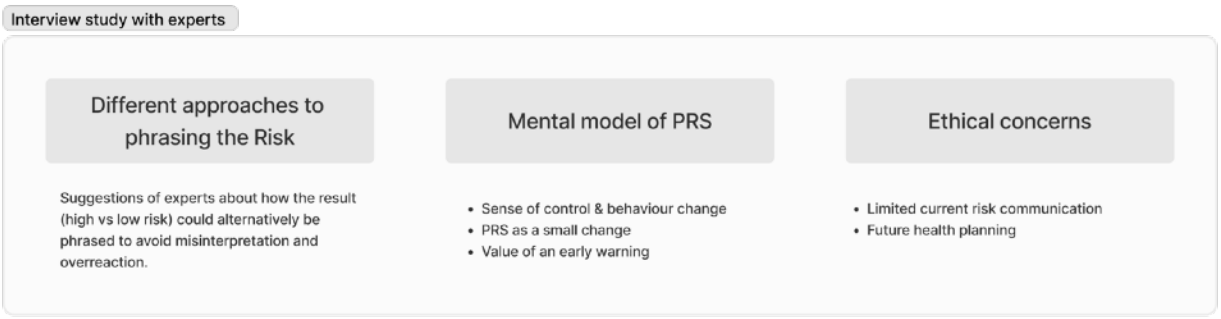


Figure 9: Overview of the themes and sub-themes identified from the expert interview study.

This chapter highlights the results from the interview study with the three experts (Figure 9). Three main themes were identified: suggestions on how to phrase polygenic risk, how to create a mental model about PRS, and ethical concerns when communicating PRS. The code tree for the expert study can be found in Appendix B.

3.4.1 Different approaches to phrasing the risk

Experts provided various insights on how PRS results should be framed to enhance understanding and reduce anxiety. An expert emphasized the ideal of face-to-face consultations but acknowledged the impracticality due to personnel and funding constraints. Reminders were suggested to help individuals remember their high-risk status and adhere to healthy behaviors and regular screenings. The experts noted that some worry is normal, as it can motivate individuals to take their health more seriously. The experts noted that genetic testing requires more explanation since it is a new and relatively complex concept. They noted that preparing for a PRS test can be challenging as people lack prior experience with it.
The phrasing of risks, such as "low risk" and "high risk," can be misleading and cause unnecessary alarm if not explained properly. Experts suggested replacing the term “high risk” with “heightened risk” to reduce alarm and clarify meaning. PRS results should be understandable to recipients with varying education levels, from basic education to doctorates, avoiding difficult terms and medical jargon without oversimplifying the information. Recipients need to understand that a PRS indicates a potential for disease based on statistical models and is one of many factors, including lifestyle and environment,

contributing to disease development. The communication should start broadly, then become more specific, and finally generalize again to avoid confusion.
A respondent from the user study highlighted that she would categorize herself into the "safe" category after receiving a low PRS result. However, the expert highlighted that this can be dangerous since a low-risk score does not imply that the risk of getting BC in the future is zero. Instead, using phrases like “we did not identify heightened risk” instead of “low risk” to avoid implying absolute safety. This approach highlights that the absence of identified risk does not change the recipient’s current health status. The use of color codes and number scales was discussed, with some experts discarding number scales due to their subjective nature and potential for misinterpretation.

"And again, as I said here, not high risk, but heightened[/elevated], maybe it would be better I would say not 'low risk', than to communicate but to say "we did not identify heightened[/elevated] risk".
A slight difference, but again it highlights that we have not identified, which is correct, and which perhaps then also does not highlight, that [the result is] low [and] don't worry, but exactly rather perhaps the wording is always, who will understand differently, that which rather then also again that everything remained as it was without that, as it was before this test, these recommendations." - Interviewee 11 (expert)

3.4.2 Mental model of PRS

Expert interviews revealed ways of calming down and comforting a PRS recipient to achieve what a user called “peace of mind about knowing [about genetic dispositions] earlier”. These communication strategies for creating an understanding (or "mental model") about the meaning and implications of a PRS result include highlighting the three following aspects: a sense of control with related behavior change, framing PRS as a small change for the recipient, and highlighting that PRS serves as an early warning which is valuable to the person.

Sense of control & behaviour change

The concept of sense of control is important for PRS recipients. Experts emphasized that it involves helping the individual understand that she has the ability to decide over her health outcomes despite her genetic predispositions. This can be achieved by highlighting actionable steps that can be taken to mitigate risks, such as lifestyle modifications like quitting smoking or reducing alcohol consumption. The expert from interview 6 stressed the importance of personalized advice, noting that when recipients see a clear path to reduce their risks, they are more likely to feel empowered rather than overwhelmed. The expert from interview 14 added that ongoing support and reminders are necessary to maintain these behavior changes, as initial motivation often fades out after a few months.

PRS as a small change

Communicating PRS results as a small change rather than a sudden, drastic shift in health status is another strategy for easing anxiety. The expert from interview 11 noted that it is important to frame the information in a way that the recipient understands her health has not changed overnight; rather, she is now more informed about her long-term risks. This perspective helps in reducing panic and fostering a more measured response to the information. The expert from interview 14 highlighted that this approach can make the information feel more manageable and less intimidating, allowing recipients to integrate the knowledge into their lives more smoothly and make gradual changes.

Value of an early warning

Framing PRS as a valuable early warning is another important communication strategy in creating a mental model about PRS. The expert from interview 11 described how early awareness of genetic risks allows individuals to take proactive measures well before any clinical diagnosis might occur. This foresight can lead to increased vigilance and adherence to preventive measures, contributing to better health outcomes. However, the expert from interview 6 pointed out that a high PRS can also cause temporary distress. It is normal for recipients to feel worried or sad initially, but this emotional response can be mitigated through effective communication and support. The experts agreed that acknowledging these feelings and providing a clear action plan can transform initial anxiety into gratitude for the early warning and motivation for positive change.

3.4.3 Ethical concerns

The experts further highlighted ethical concerns when communicating PRS. These relate to highlighting that PRS does not constitute the full genetic risk of a person and that it should be considered how the communication may impact a PRS recipient's future health planning.

Limited current risk communication

Experts highlight that PRS does not constitute the entire genetic risk. Although PRS shows a part of the risk, monogenic risk scores (e.g., used when finding inherited genetic mutations in family situations) can add an additional perspective, contributing to the comprehensiveness of genetic risk. In turn, considering that EHIF's personalized screening program only bases its results on PRS, this should be reflected in the communication of the results. This is so that recipients wouldn't mistake the result for a full risk overview. All three experts consider that there should be more genetic risk communication to families.

The expert further highlighted that the model accuracy should be included. This relates to the genetic population for which the model predicts risk most accurately. Since the PRS model of the Estonian Biobank was reported to be trained on people with Estonian heritage, this limitation should be communicated. Nonetheless, according to the expert, non-genetic risk can be communicated to non-ethnic Estonians. It should also be mentioned that the result is based on current medical knowledge and may change in the future.
Another expert noted that the focus of the PRS communication should be on the follow-up steps, and not how high the risk is. The expert highlighted that the risk level and related statistics can anyways be interpreted differently by every recipient and one should shift the focus onto emphasizing concrete follow-up steps.
Furthermore, educational support alongside the PRS result was deemed important by respondents. Proper information materials should be provided, explaining what the PRS entails, how the results were derived, and the actual meaning of terms like "risk." This helps in setting realistic expectations and reducing unnecessary panic. Furthermore, the information materials should be of high quality.

"Surely it is possible, surely these information materials would be needed. On the one hand, so that health professionals know, but on the other hand, so that the general public itself knows, all kinds of introductory videos, well, perhaps leaflets are not enough, but perhaps we also need to put some kind of introductory videos out there, somewhere on YouTube, about what exactly we are doing, what this result means, in what cases it is not advisable. Exactly that if you have in your family, for example, here the mother has breast cancer under the age of 50, or a sister or someone close to the family, that you should still look at the fact that a consultation with a medical geneticist is also recommended." - Interviewee 14 (expert)

Future health planning

This involves discussions about the implications of the genetic risk for family planning and the potential risks for children or other family members. Genetic counselors often play a crucial role in providing this type of advice, helping patients understand the inheritance patterns and possible preventive measures for their descendants. According to the experts, women may seek a healthcare provider's expertise to discuss how the results might affect their long-term health and to verify the accuracy and implications of their PRS results.

"... but there are so many ethical problems that we run into when we communicate genetic information. First of all, how will this affect my children? How does it affect me because risk is just risk. Even if I say your risk of getting breast cancer is 80%, it's actually 20% of not getting it in your lifetime. But how is that person going to make choices in their lifetime? Given their genetic information." - Interviewee 6 (expert)

Conclusion on the expert study

The expert study revealed insights into the communication of PRS for BC. Experts highlighted the importance of clear, non-alarming language, preferring terms like “heightened risk” over “high risk” to reduce anxiety. The communication should avoid medical jargon while remaining informative. Communicating PRS involves emphasizing actionable steps recipients can take, such as lifestyle changes, to manage their risk. PRS should be presented as an early warning that encourages proactive measures, with ongoing support to maintain behavior changes. Ethical considerations were highlighted, ensuring recipients understand the limitations and accuracy of PRS, especially across different genetic populations. Lastly, communication should support informed health decisions and consider the impact on the recipient's future health planning.

3.5 Discussion

The findings from the interview studies with potential users and experts provide comprehensive insights into the needs and concerns related to PRS communication for BC. These findings show important implications for the design of a digital communication tool. Users reported feeling overwhelmed by the information provided in the PRS, leading to anxiety and distress. This emotional response was particularly strong when the PRS indicated a high risk. The design should prioritize clarity and simplicity in the presentation of PRS results. This can be achieved by summarizing key information upfront and using visual aids such as color codes and simple charts to convey the risk levels.

Additionally, providing immediate access to support, either through a digital chat function or a hotline, can help users manage their initial emotional responses. Many users confused the PRS with a definitive diagnosis, which could lead to unnecessary panic and misinformed decisions. The communication tool must clearly differentiate between risk and diagnosis. This can be done through explanatory sections that detail what a PRS indicates and what it does not. Using phrases like "heightened risk" instead of "high risk" can help mitigate misunderstandings. Including educational resources such as videos and FAQs can further aid in clarifying these concepts.

Users expressed a strong desire for practical advice and follow-up steps after receiving their PRS results. This includes lifestyle recommendations and tailored screening plans. The digital tool should offer personalized action plans based on the PRS results. This could involve integrating algorithms that provide tailored lifestyle recommendations and scheduling reminders for follow-up screenings. Ensuring that users have easy access to their health history and upcoming appointments through the tool can also enhance its usefulness. There was a preference for face-to-face or at least video consultations when dealing with high-risk results, as these interactions are perceived to provide a more supportive and personalized experience.

While the initial communication can be digital, the tool should facilitate easy scheduling of in-person or video consultations with genetic counselors or healthcare providers. This hybrid approach can combine the efficiency of digital tools with the empathy and personalized support of human interactions. Users need help developing a mental model of what PRS means and how it fits into their overall health context. This includes understanding the implications of their results and how to manage their risk. The tool should include comprehensive educational content that helps users build a mental model of PRS. This can be achieved through interactive tutorials, explanatory videos, and real-life examples of managing genetic risk. Providing a narrative that places PRS within the broader context of the user's health can help make the information more relatable and less abstract.

Trust in the institution behind the PRS communication and concerns about data privacy were significant for users. The design should ensure transparency about data usage and privacy. Clear statements about how user data will be handled, stored, and protected should be prominently displayed. Building a reputable brand for the tool, possibly through partnerships with recognized healthcare institutions, could also enhance trust.

The insights from the interview studies directly inform the design of the digital communication tool for PRS. To meet the informational and social support needs of the target group extracted from the interview studies, the conceptual design should focus on the elements highlighted in Table 5.

Considerations for the design process from literature and the interview studies	**Explanation**
Clear and simple presentation	Use intuitive visual aids and straightforward language to present PRS results, avoiding medical jargon
Immediate support	Include features for immediate access to support, such as chat functions with genetic counselors or helplines
Personalized action plans	Offer tailored lifestyle recommendations and follow-up plans based on individual PRS results
Hybrid communication channels	Facilitate both digital and in-person consultations to cater to varying user preferences, especially for high-risk communications
Comprehensive educational resources	Provide interactive and multimedia educational content to help users understand and manage their genetic risk
Trust and transparency	Ensure transparency in data handling and building trust through reputable partnerships and clear communication about data privacy

Table 5: Considerations for the design process, extracted from the results of the user and expert interview studies.

3.6 Overall conclusion of the user and expert studies

The results chapter of this study addresses the research question: "What informational and social support do women receiving a personalized risk score for breast cancer need in the context of a digital communication tool?" Through user and expert interviews, several key themes emerged that directly inform the design of an effective digital communication tool. Participants often felt overwhelmed by the PRS information, leading to anxiety and distress. This underscores the need for clear, simple presentation of information, using visual aids, and avoiding medical jargon. Many participants also confused PRS with a diagnosis, highlighting the importance of clearly differentiating between risk and diagnosis, using terms like "heightened risk" instead of "high risk." Users expressed a strong desire for practical advice and follow-up steps, including tailored lifestyle recommendations and screening plans. This indicates that the tool should offer tailored action plans based on PRS results, integrating algorithms to provide tailored recommendations and reminders for follow-up screenings. Additionally, high-risk results should be communicated through face-to-face or video consultations to provide personalized support, while digital communication suffices for low-risk results.
The need for comprehensive educational content was also evident, as users require help building a mental model of PRS and understanding its implications. Trust and data privacy are significant concerns; Thus, transparency in data usage and building a reputable brand through partnerships with recognized healthcare institutions are important.

Extracting insights for the design process from these findings, the digital communication tool should:

- Use clear, simple language and visual aids to present PRS results,
- include immediate support features, such as chat functions with genetic counselors or helplines,
- offer personalized action plans and follow-up steps based on individual PRS results,
- facilitate both digital and in-person consultations, particularly for high-risk communications,
- provide comprehensive educational resources to help users understand and manage their genetic risk, and
- ensure transparency in data handling and build trust through reputable partnerships.

4. Conceptualization

This chapter presents the conceptualization stage of this thesis. To arrive at the deliverables, which are a service blueprint and roadmap, multiple activities were performed. First, a persona technique was used to capture specific user values but use them as the basis for the following design process. Secondly, based on the results chapter, an early exploration was done into the following design process. Thirdly, a design goal was formulated, along with associated design criteria for the design concept, and a How Might We statement. In the following chapter, the service concept is introduced along with the service blueprint. Afterwards, the design roadmapping process is described, highlighting the resulting strategic and tactical roadmaps.

4.1 Persona technique

4.2 Early directions for the following design process

4.3 Design goal

4.4 Design criteria

4.5 How Might We statement

4.6 Service concept

4.7 Roadmapping

4.1 Persona technique

To formulate an overview of the different types of users and their needs, the persona technique guided by Miaskiewicz & Kozar (2011) was used to qualitatively formulate personas based on the interview respondents. Here, qualitative summary notes from the interviews were combined with assistance from ChatGPT for clustering. This resulted in the following personas, highlighted in Figure 10.

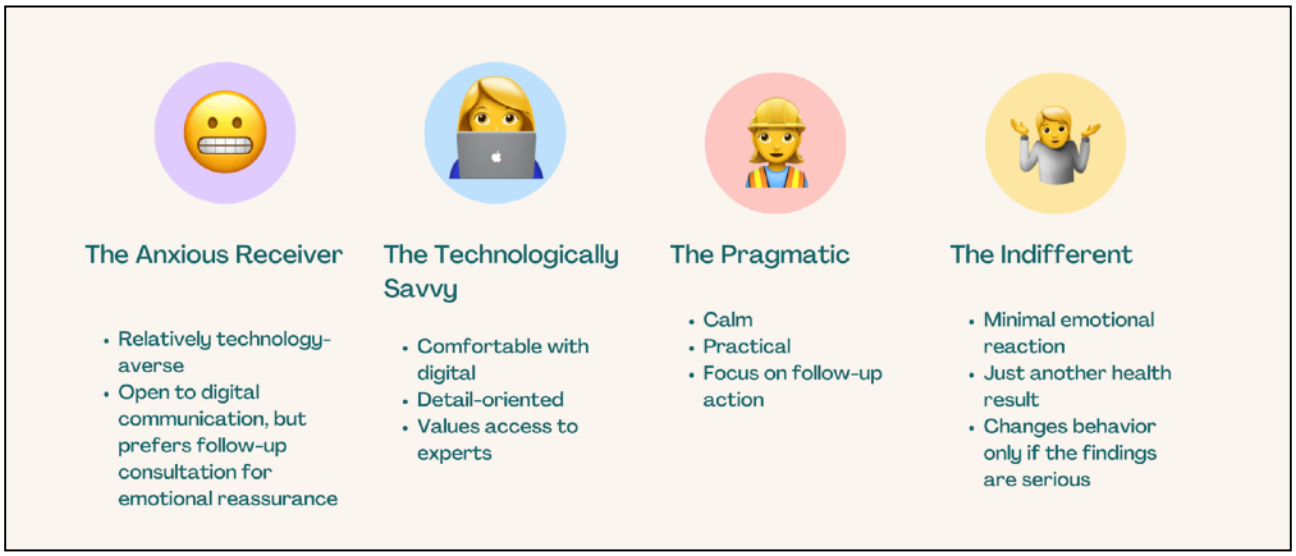


Figure 10: The four personas with their main characteristics.

1) The Anxious Receiver

This persona was based on interviews 1, 2, 5, and 9. The Anxious Receiver is primarily driven by anxiety when receiving medical results, particularly those related to PRS. This persona's primary concern is understanding the implications of a high score and mitigating potential health risks through detailed consultations and continuous support. She is used to receiving the PRS result online, along with other medical results. She prefers to have a first look at the result on her own, think of questions for the doctor, and then reach out. If invited to the doctor's without prior knowledge of the score, she will interpret it with further anxiety, assuming the result is highly alarming.

Continuous support through regular follow-ups and the availability of medical professionals for additional questions is important to her feeling secure and informed. She is cautious and detail-oriented, often overthinking potential health issues and preferring to have all possible information and scenarios laid out before her. She is proactive about her health due to the experiences of friends or distant relatives. She is routine-oriented, scheduling regular health check-ups and screenings.

She is relatively technology-averse and prefers in-person consultations over digital communications for critical health information, highlighting her need for personal and empathetic interactions. Her reaction is characterized by worry and a need for immediate reassurance. As a proactive information seeker, once a health concern arises, she actively seeks information from credible sources and professionals to comprehensively understand her health situation. Her emotional dependency often makes her feel overwhelmed by potential health risks, seeking comfort from healthcare professionals who can provide clear explanations and reassurance. She likely seeks second opinions from further health professionals.
She primarily reaches out to her GP as the first point of contact, providing initial consultations and referrals. She considers nurse practitioners easily accessible for quick consultations and to bridge the communication between the patient and the GP. She may also consult with a specialist, such as a genetic counselor, for expert advice and detailed risk assessments.

Reactions to a low PRS result

The Anxious Receiver would initially feel relieved but would still seek reassurance from healthcare professionals to confirm that the low score indicates low risk. She would likely schedule an appointment with her general practitioner to discuss the results in detail. Despite the low score, she would continue her routine health check-ups and screenings, as her anxiety would drive her to stay careful about BC risk.

Reactions to a high PRS result

Receiving a high PRS score would trigger significant anxiety and concern. She would immediately seek a detailed explanation of what the high score means for her health and what steps she can take to mitigate the risk. She would prefer in-person consultations to digital communications, valuing interactions that help her feel more secure. She would likely aim to gather as much information as possible and to receive a comprehensive action plan.

Unique characteristics of the Anxious Receiver include a strong need for reassurance due to her anxiety about health, making her reliant on medical professionals to mitigate her fears via thorough explanations and reassurance. She has a preference for in-person consultations over digital ones, (despite the convenience of digital results) due to the personalized interaction and the possibility to ask immediate follow-up questions.

2) The Technologically Savvy

The Technologically Savvy was based on interviews 3, 8, and 10. This persona represents individuals who are comfortable with digital technology and prefer using online resources for accessing and managing their health information. She is highly educated, tech-savvy, proactive, independent, and detail-oriented. She regularly uses digital devices, including her computer and smartphone, to manage daily tasks. She is health-conscious, prioritizes her well-being, and integrates new health insights into her lifestyle, preferring self-service options for scheduling appointments and accessing health records.

She is comfortable receiving PRS results and other medical information via email or the Health Portal. She expects clear, detailed explanations of these results, including the next steps. While she prefers digital communication, she values having the option to access to medical professionals for follow-up questions or concerns, if necessary, and does not mind telehealth consultations.

As a first general point of contact in case of medical questions, she typically consults her general practitioner or a specialist for detailed discussions. She prefers consulting professionals who are knowledgeable and can provide in-depth explanations about complex medical information. Unique characteristics of this persona include extensive integration of technology into her health management routines, looking up clarifications about medical results online, and valuing quick and easy access to health information through comprehensive and user-friendly digital platforms.

Reactions to a low PRS result

The Technologically Savvy would react positively to a low PRS score, feeling reassured by the favorable result. She would likely access her results via the Health Portal and might google to confirm the implications of a low score. She would appreciate concise and clear digital explanations and would not feel the need for extensive follow-up consultations unless she had specific questions.

Reactions to a high PRS result

A high PRS result would prompt her to seek detailed information online first, using reliable health websites and forums to understand the implications. She would value detailed, yet concise digital explanations and might schedule a telehealth consultation with her GP or a specialist for further clarification. While she is comfortable with digital communications, she would ensure that she has access to expert advice and would use online tools to manage her follow-up actions efficiently. She would integrate recommended lifestyle changes into her routine and might use health apps to monitor her progress.

In contrast to the Anxious Receiver, the Technologically Savvy prefers detailed descriptions that are concise, straightforward, and accessible online. She appreciates having the option to

delve into more detailed information if needed but prefers initial explanations to be brief and to the point. She values the convenience of accessing this information digitally and uses online resources to seek additional clarification before consulting a healthcare professional.

3) The Pragmatic

This persona was based on interviews 4, 7, 12, and 15. The Pragmatic is defined by a practical and solution-oriented approach to health information. She is typically well-organized, balancing multiple responsibilities in her professional and personal life, including family activities and maintaining a healthy lifestyle. She values clarity and efficiency in communication and is career-oriented, often holding a position that requires interaction with various stakeholders such as government entities, organizations, or clients.

She is comfortable with digital tools and e-services, which she uses regularly for both work and personal matters. Her pragmatic mindset means she prefers straightforward, reliable information and tends to avoid overly emotional or speculative discussions. She is proactive about health management, integrating preventive measures and regular check-ups into her routine.

When receiving PRS results or other medical information, she focuses on understanding the practical implications. She views PRS as a tool for informed decision-making rather than a definitive diagnosis. She expects clear, concise information that outlines the next steps and practical actions she can take to manage her health.

Her emotional support needs include reassurance that the information she receives is accurate and actionable. She prefers calm and collected communication from healthcare providers to help maintain her calm attitude. Additionally, she values being in control of her health information and decisions, which minimizes unnecessary anxiety by focusing on factual information. Her informational support needs include clear explanations of what her PRS results mean, including any potential health risks and recommended actions. She expects detailed steps she can take to mitigate risks or improve her health, integrating these into her routine efficiently. She trusts information from established, credible sources and appreciates references to supporting data or research.

Reactions to a low PRS result

The Pragmatic would view a low PRS score as a positive affirmation of her current health management strategies. She would appreciate the clear and concise communication of the results and would integrate this information into her routine without much emotional reaction. She might consult her GP for a brief confirmation but would not feel the need for extensive follow-ups. Her focus would be on maintaining her healthy lifestyle and continuing her preventive measures.

Reactions to a high PRS result

Upon receiving a high PRS score, the Pragmatic would seek to understand the practical implications and the actionable steps she can take. She would prefer straightforward and reliable information from credible sources. A high score without clarification may prompt her to consult with her GP and possibly a specialist to discuss preventive measures and risk mitigation strategies. She would integrate the recommended actions into her daily routine efficiently, focusing on practical and effective health management without succumbing to unnecessary anxiety.

Her typical first contact in case of health questions or concerns is her GP or a specialist relevant to the health concern. She values the opinion of experts who can provide concrete advice. She prefers consultations that are to the point, focused on practical advice, and free from unnecessary technical jargon, unless it is explained in an understandable manner.

4) The Indifferent

This persona was based on interview 13. The Indifferent persona shows minimal emotional response and treats the PRS result as just another piece of health information. She does not significantly alter her behavior unless presented with substantial findings. She approaches health information with a logical mindset, weighing the practical implications rather than emotional reactions. While she pays attention to her health, she does not overemphasize individual pieces of information unless they indicate a clear need for action. She values autonomy in making health-related decisions and prefers having control over her health information.

In terms of lifestyle, she is likely to have a demanding job and multiple responsibilities, requiring efficiency in managing health-related matters. She is comfortable using digital tools for managing health information, such as accessing results online or using digital health platforms. She prefers regular check-ups and screenings but does not worry about potential health risks unless they are imminent.

When receiving a PRS result, she does not experience significant anxiety or stress. She favors receiving and reviewing health information through digital means rather than in-person consultations unless strictly necessary. She prefers brief and to-the-point explanations of her PRS results without unnecessary details. She utilizes online platforms and digital tools to schedule appointments, access medical records, and receive test results. She maintains a balanced view of health risks, neither underestimating nor overreacting to potential health issues. She is capable of making quick decisions regarding health management, relying on clear and actionable information. Although she remains relatively detached emotionally, she takes responsible actions based on the information provided, ensuring her health is adequately monitored and managed.

Reactions to a low PRS result

The Indifferent persona would likely take a low PRS result in stride, viewing it as just another piece of information. She would not alter her behavior significantly unless prompted by her GP or if the information indicates a clear need for action. She might glance over the digital results and appreciate the brief, to-the-point explanations, but would not seek additional consultations unless absolutely necessary.

Reactions to a high PRS result

A high PRS result would not elicit a significant emotional response from the Indifferent persona. She would logically evaluate the information and decide on follow-up actions based on the practical implications. If the high score indicates a substantial risk, she would contact her GP for a brief consultation to understand the necessary steps. She values efficiency and would use digital tools to manage her health information, schedule appointments, and access records online. She would only seek further consultations if the high score necessitates a change in her health management strategy.

To further contrast the Pragmatic and the Indifferent, the Pragmatic is action-oriented and focuses on the next steps, while the Indifferent is efficient in decision-making but only takes action if there's a clear need. The Pragmatic requires detailed explanations of the next steps, while the Indifferent prefers concise information and conditional support based on the severity of the findings. Table 6 summarizes how each persona would react to a low and a high PRS result. Figure 11 maps the four personas onto different axes for comparison in relation to tailoring digital PRS communication for them.

Persona	Reaction to low PRS	Reaction to high PRS
Anxious Receiver	Initially relieved but would still seek reassurance from healthcare professionals	A high PRS may cause anxiety and concern; Prefers to get to know the result online and then reach out to a healthcare professional for a consultation at the earliest convenience
Technologically Savvy	Feeling reassured; Looking for concise and clear digital explanation (including online sources); would reach out to a healthcare professional only in case of specific questions	May want to contact experts if no clarification is given about the implications of a high PRS result
Pragmatic	Views low PRS as affirmation as positive affirmation of her health lifestyle choices	Without further information, a high PRS may prompt her to consult with her GP or other healthcare specialist to discuss preventive measures and risk minimization

Indifferent	Would react calmly to a low PRS; Would not change behaviour unless directly prompted to; Would not seek additional consultations unless absolutely necessary	Would evaluate the information logically; Would only seek further (short) consultations if the high PRS requires a behavior change

Table 6: Summary of the reactions to a PRS result by risk level and persona.

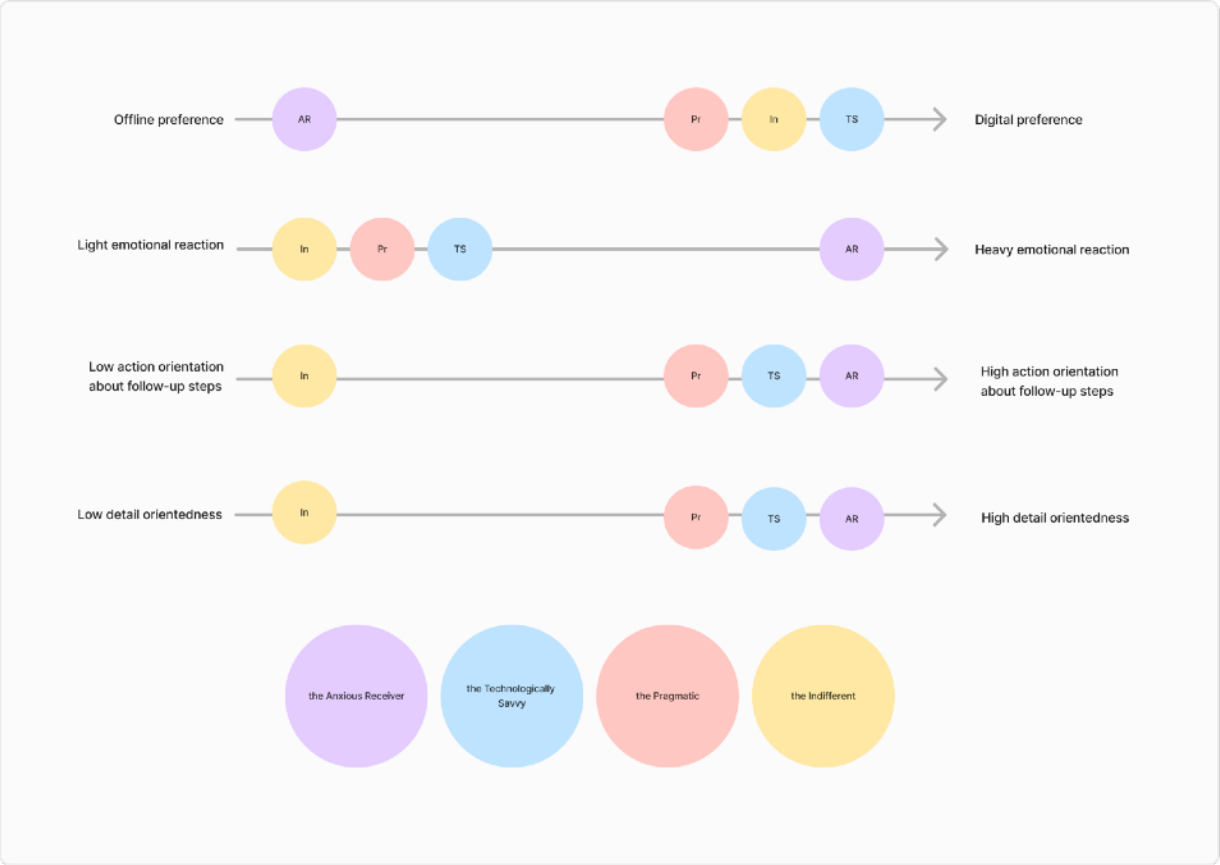


Figure 11: The four personas plotted on four different axes: online vs offline preference, light vs heavy emotional reaction, low vs high action orientation about follow-up steps, low vs high detailed orientation

The persona technique highlights that all four personas are open to receiving the initial PRS result via the Health Portal since they are used to being communicated medical results online. However, their behavior after the result communication differs. The Anxious Receiver would most likely expect a consultation with a healthcare professional at the earliest convenience. The Technologically Savvy and the Pragmatic may not necessarily reach out to a healthcare professional, but they appreciate the option to do so. The Indifferent is the least likely to reach out to a healthcare professional (in case of either low or high PRS), only doing so when the situation is perceived as serious and immediate intervention is necessary.

The results from the persona technique relate to findings from related academic research into patient profiles (Groeneveld et al., 2019; Figure 12). The Anxious Receiver aligns closely with the Managing Profile due to the high need for open and emotionally supportive

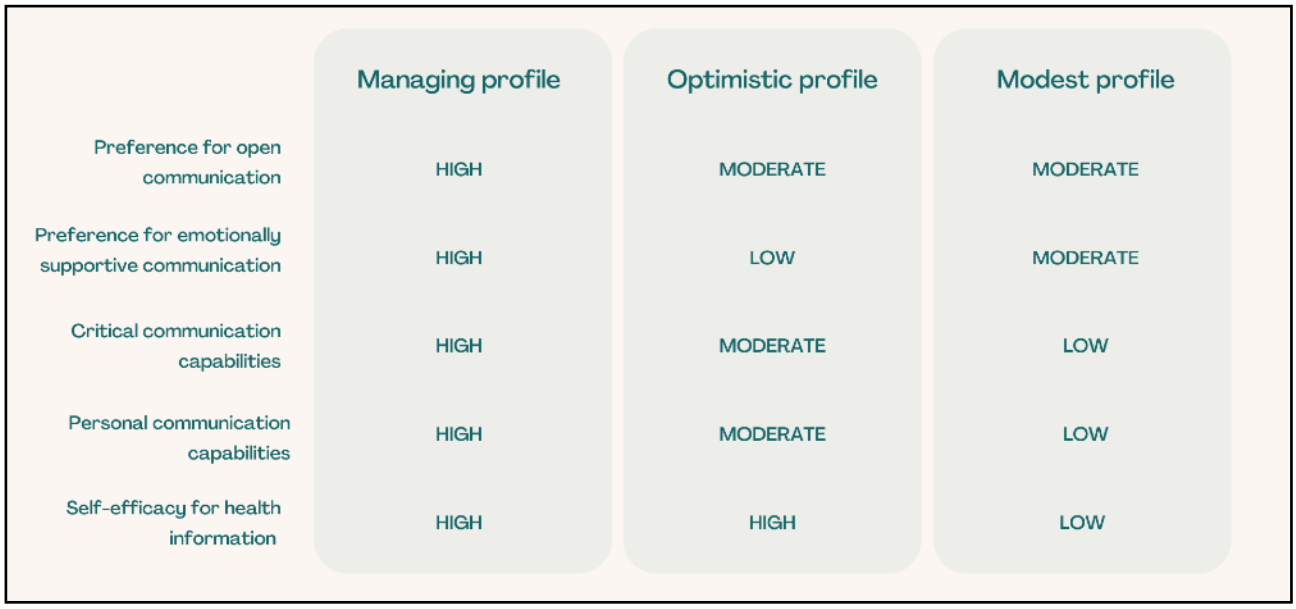


Figure 12: The three patient profiles according to Groeneveld et al. (2019)

communication, requiring detailed consultations and continuous support. The Technologically Savvy fits the Optimistic Profile, characterized by a moderate preference for open communication and high self-efficacy for health information, preferring concise, clear digital explanations. The Pragmatic also aligns with the Optimistic Profile due to her preference for practical and straightforward information. Lastly, the Indifferent persona corresponds with the Modest Profile, which has low critical and personal communication capabilities and low self-efficacy for health information, showing only a moderate preference for open communication and taking action only when there is a clear need.
However, there are critical differences that do not overlap between the personas and Groeneveld et al. (2019) profiles. The Anxious Receiver might exhibit a higher level of dependency on healthcare professionals than the Managing Profile, indicating a potential gap in self-efficacy for health information. The Technologically Savvy persona's reliance on digital tools might not be fully captured by the Optimistic Profile, which might encompass a broader range of communication preferences. The Pragmatic persona's emphasis on efficiency and practicality might not align entirely with the moderate critical communication capabilities of the Optimistic Profile. Finally, the Indifferent persona may display a higher level of disengagement than the Modest Profile, potentially underestimating the latter's moderate preference for open communication.

4.2 Early directions for the following design process

Driven by insights from the results section and the persona method, an early exploration was done into possible directions for the design process. The mind-mapping method (Van Boeijen et al., 2014) was used to brainstorm early design directions. The starting node was called

"Avoiding overreaction after digital PRS communication". This was further elaborated regarding the design context, the behavior of PRS recipients after the result communication, and the media for interaction. As a result, the following ideas arose: spreading out the information delivery throughout the user journey, designing for perception change of the service, use of an AI chatbot to emulate an interaction with a genetic counselor. These ideas relate to early directions for the following design process, which are refined and elaborated in chapter 4.6.

1. Overwhelming delivery of PRS results

Multiple interview respondents highlighted the report as potentially causing anxiety, stress, and overreaction. During the mind-mapping, it was considered that previously it had been assumed that the recipient gets the majority of information about the genetic test after the blood sample has been given. Thus, considering that the potential users from the interview study currently have relatively little awareness and knowledge about genetic testing, a detailed and comprehensive result communication may actually overwhelm the respondents. To combat this, the information delivery can be spread out over the user journey. For example, the information about the PRS can be moved before results delivery.
Taking into account insights from the results section, to avoid an overwhelming delivery, the information needs to be presented in a clear and simple format, with the possibility to read more into the topics independently. Moreover, there should be an action plan presented to the recipient about follow-up steps. Interviewee 14 (expert) highlighted that the level of risk (e.g., "high" vs "low" risk) can be easily interpreted differently by a person. Thus, to avoid misinterpretation, the focus can instead be put on what the follow-up steps are post-result. To clarify the information, immediate support should be available to support the user. Users wishing to consult a healthcare professional in person should be presented with a way to do so.

2. Perception change of the service

Insights from the results section that the service under the name "personalized breast cancer screening program" can be misleading and confusing to potential users. The result can be mistaken as a diagnosis and the recipients may connect the service with characteristics typical of screening programs, such as the use of mammography. To change this potential perception, the presentation of the service should be altered. As examples, a branding strategy and associated marketing campaigns were considered.

3. Early exploration into AI as a tool for communicating PRS

Insights from the results section include PRS recipients wishing for immediate support in handling their health situation. However, the Estonian healthcare system faces a personnel shortage in the future (ERR, 2024). Thus, AI tools were considered as a future opportunity to offer immediate support without necessary human intervention.
A respective exploration was done into using AI tools for communicating PRS. First, AI chatbots were considered a relatively simple and accessible solution for answering follow-up questions. Large-language models (LLMs) like OpenAI's ChatGPT, Google's Gemini, and Meta AI have several advantages, including round-the-clock availability and enhanced accessibility (Dimitriadis, 2020). These chatbots can provide immediate, personalized advice based on user input, which is particularly useful for follow-up questions. Recent advancements have made these interactions more human-like, increasing their perceived intelligence and usefulness (Awasthi & Kaveri, 2023). Customization features in ChatGPT, for example, allow for the creation of tailored chatbots that can offer personalized follow-up support based on the specific needs and information provided by the user (OpenAI, 2023).
Secondly, in a more disruptive and future-oriented exploration, the potential role of AI as digital genetic counselors was considered. The idea is rooted in interview respondents mentioning a wish for human interaction, and this exploration into AI agents designed to emulate human touch (Soul Machines, 2022) serves as a potential contender. Thus, the concept goes beyond simple follow-up questions, envisioning AI agents partially replacing the role of human genetic counselors. These AI-driven virtual assistants, specialized in genetic counseling, could handle initial PRS communications, determine which individuals need to be referred to human genetic counselors, and provide ongoing support. This approach utilizes the advanced capabilities of LLMs to deliver personalized and context-aware advice, ensuring that users receive accurate and timely information while aiming to emulate a certain degree of human touch in the communication process.

This early exploration into possible design concepts helped to come up with the design goal, design criteria, and a How Might We statement. They are introduced in the following.

4.3 Design goal

Based on the original design brief and the findings from the user and expert study, the general objective for the design process was clarified:

> The overarching design goal is to reduce the proportion of women reaching out to a medical professional after getting invited to the

personalized screening or after receiving their PRS result, via a digital communication tool.

To formulate the design goal into a specific How Might We statement, I summarized (according to the results from both the user interviews as well as the persona technique) why women tend to reach out to a medical professional after digital BC PRS delivery.
Women tend to reach out to a medical professional...

- to ask for follow-up questions and clarification, including personalized explanations, and
- to receive emotional support to handle the potential anxiety and distress arising from interpreting the result, also including overreaction about overstating the implications of the result (e.g., by mistaking the risk for a diagnosis).

These aspects relate to feelings of confusion, concern, anxiety, and shock. To address these emotional reactions in the design intervention, process behaviors and cognitive strategies (methods) that genetic counselors use to communicate genetic risk were summarized in Table 7 from both the literature review and the user study.

Method	**Explanation**	**Source**
Empathic statements	The genetic counselor interacting with the counselee in an emphatic way	Gupta et al. (2021); Lobb et al., (2005)
Paraphrasing and summarizing (Reflection)	Processing information by paraphrasing and summarizing to give reassurance and validation	Gupta et al. (2021)
Clarification	Clarifying misunderstandings and explaining medical terms	Gupta et al. (2021)
Analogies	Using analogies to explain the PRS, including visual aids	Gupta et al. (2021)
Giving a sense of control	Providing the recipient with a sense of control to highlight the value of behavior change.	Expert interviews
Framing genetic risk as a small change	Focusing on genetic risk as a small change, in that the PRS result does not imply an immediate threat to the person.	Expert interviews
Value of an early warning	The person should understand that it is a positive thing that she got to know about the risk early on, so that risk of future disease development can be minimized.	Expert interviews; Gorman et al. (2022)

Table 7. A summary of different process behaviours and cognitive strategies used by genetic counselors.

It was decided that the service concept would serve as a tool for reducing anxiety, stress, and worry for PRS recipients. All personas are open to receiving PRS results online (as indicated by the user study and persona technique), with the Anxious Receiver persona most likely to reach

out to a medical professional. Thus, in the following design process, it was considered that the digital PRS communication tool should focus on providing reassurance to all personas but would take into account that the Anxious Receiver expects a consultation after the digital communication.

Taking into account the insights from the user study, I decided that the PRS result would be communicated online for all personas. Insights from the results section highlight that going to a doctor's office to communicate the score in person may have the opposite effect. Since the interview participants are used to digital communication of health results, they may interpret an invite to a PRS consultation as a potential sign that the result is significantly problematic, resulting in unnecessarily increased anxiety and worry. Thus, it was decided that the PRS result would be communicated online, with the option to reach out to a medical professional afterward.

4.4 Design criteria

To reduce the proportion of women contacting a medical professional after PRS receipt, it is important to ensure that recipients do not overreact to the result. The insights from the literature review, user interviews, and genetic counselors' methods in consulting PRS recipients were formulated into the following design criteria (followed by the source of each criterion). The communication should...

1. be easy to understand for a variety of numeracy levels (literature review, user study, expert study).
2. highlight that PRS is a small change for the recipient (user & expert study).
3. that the communication serves as a valuable early warning (expert study).
4. highlight the health-related implications for the recipient (user study).
5. highlight the follow-up plan and that the person has a sense of control in managing their risk (user study & expert study).
6. highlight the limitations of the result (expert study).
7. be able to adjust as the overall service develops further (EHIF’s future plans).

4.5 How Might We statement

The conceptualization section of this thesis is based on the following assumption: If women get peace of mind about their genetic health, they are less likely to contact a medical professional before or after receiving their PRS score. Using the insights from the results section, peace of mind is defined as the PRS recipient understanding the meaning and implications of genetic risk in a way that keeps the participant calm and reassured.

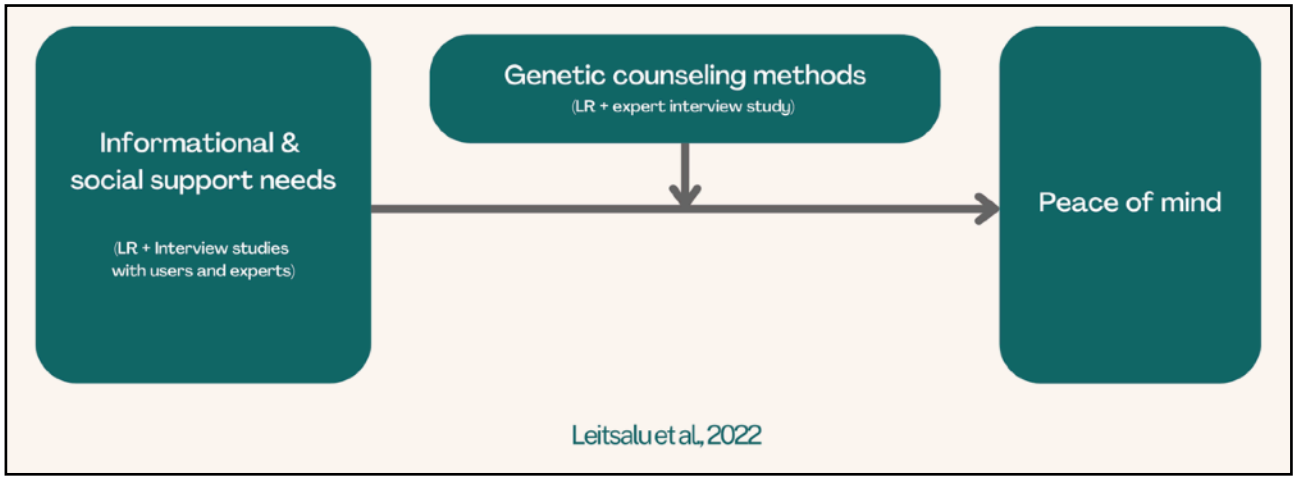


Figure 13: A schematic showing the approach to creating a peace of mind for PRS recipients by responding to informational and social support needs by utilizing genetic counseling methods (Leitsalu et al., 2022).

Since recipients with a high PRS experience the most anxiety, stress, and worry, the following service concept focuses on communicating high PRS to have the biggest impact on reducing potential strain on the healthcare system due to women reaching out to consultations. Based on the design brief, the user study, the personas, and the different genetic counseling techniques, the following How Might We (HMW) statement (IxDF, 2016) was formulated:

> How might we use genetic counseling techniques to create peace of mind for female recipients of a high PRS result?

Thus, the following design process aims to communicate BC PRS by responding to informational and social support needs (identified from the literature review and the interview studies with both users and experts) by using genetic counseling methods (identified from the literature review and the expert interview study) via a digital communication service (Figure 13).

4.6 Service concept

The resulting concept is called My Genetic Mirror (overview in Figure 14). It is a sub-page on the Health Portal that aims to communicate an individual's genetic risk by formulating it as a distinct type of medical result. This is achieved by a branding concept to distance the service perception from typical health results, and an interactive fire safety metaphor to explain the PRS result in a simplified way. The metaphor enables the recipient to imagine genetic risk as the future risk of having a fire at one's home, and that the subsequent BC screening program

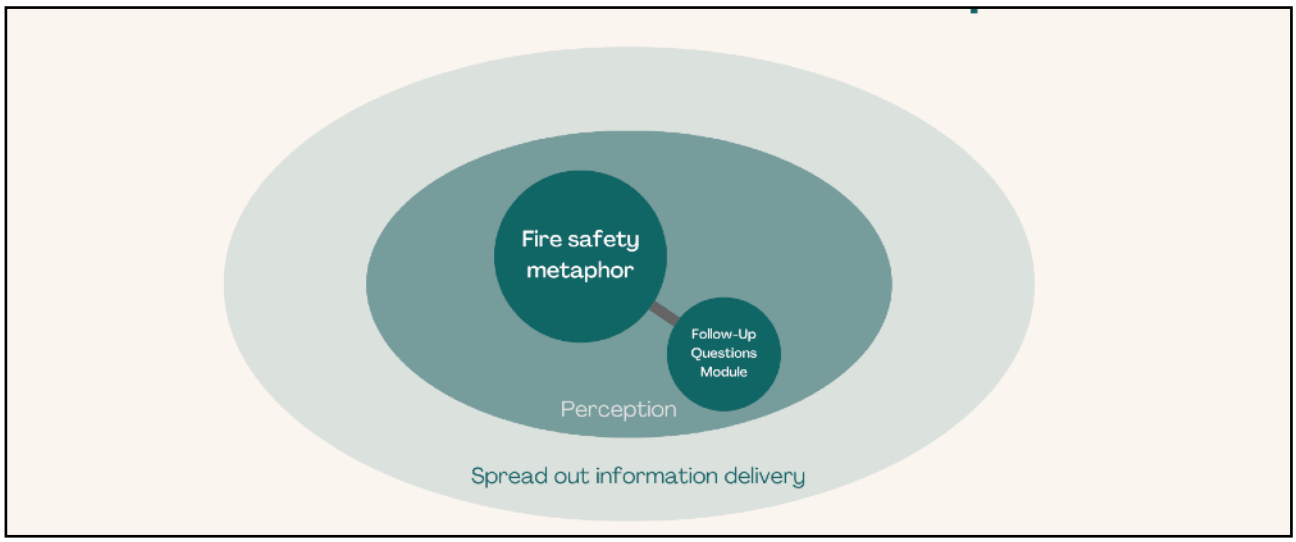


Figure 14: Overview of the My Genetic Mirror concept. The core of the concept is made up by a fire safety metaphor, along with a Follow-Up Questions Module. These two are presented to the users via the branding concept, which makes up the perception of the service. The outer layer of the service constitutes that the information in the My Genetic Mirror concept is introduced in a spread-out manner to avoid overreaction.

(in case of high risk) is like a smoke detector helping to identify danger early. The user needs and genetic counseling methods incorporated into the service are highlighted in Tables 8 and 9. Furthermore, a Follow-Up Questions Module is included to provide answers to PRS recipients' questions and concerns. To avoid overreaction when receiving a PRS score, the information delivery is spread out.

The design concept is formulated on two different levels: the service blueprint and two roadmaps. The service blueprint provides an insight into how the website, along with street advertisements and email invites, onboards the user into the program and subsequently communicates the PRS result with a potential follow-up action plan. The roadmaps were made with the future perspective in mind – taking into account user values, trends, technologies (among others) and how should the service proposition be developed further during the next five years.

The different elements of the service and its future development were conceptualized based on the needs and wants of the personas. Figure 15 shows which personas each element focuses on. In the following, an overview of the roles of each service component is given.

Spreading out information delivery

This element focuses on avoiding overwhelming the PRS recipient with information by introducing the concept of genetic risk and the service step-by-step. Its aim is to first communicate to the recipient the concept of the service, its outcomes, and implications before the actual result is communicated.

Figure 15: Overview of which personas are targeted the different service concept elements.

Branding

The aim of the branding is to change the perception of the service to less "medically threatening". Since the users may consider the PRS result as directly affecting the person's health, the branding approach aims to avoid users overstating the implications of the result, and thus preemptively calm them down.

Fire safety metaphor

Related to the branding concept, the metaphor also aims to preemptively calm down the users. Whereas the branding concept focuses on the general (first) impression of the service, the metaphor aims to help create a mental model in the mind of the recipient to imagine the meaning and implications of the service. For this, genetic risk and the service are paralleled with a fire safety metaphor.

Follow-up Questions Module (FUQM)

The FUQM serves as the initial touchpoint for getting answers to general and specific questions about the result. It includes an frequently asked question (FAQ) section for general questions, a human-powered chatbot for more specific questions, and contacts to healthcare professionals for a more thorough interaction. It aims to get the most pressing questions answered right away, helping the recipient to understand her situation and determine, whether a follow-up consultation is necessary. There, the chatbot serves an initial triage function, inviting those with a more urgent situation for a consultation.

User need for digital PRS communication (from the literature review and interview study)	**Application in the design solution**
Clear and simple presentation	The PRS result is formulated in an concise way, assisted by a fire safety metaphor to simplify the concept of genetic risk for the recipient.
Immediate support	Chatbot function for follow-up questions and clarifications
Personalized action plans	The PRS result is formulated in an actionable way, immediately highlighting the what the person should do to minimize her risk. The fire safety metaphor simplifies and summarizes what the recipient should do as follow-up steps.
Hybrid communication channels	The service concept includes the option to schedule a consultation with a healthcare professional, should the digital communication not suffice (mainly intended for the Anxious Receiver personality).
Comprehensive educational resources	The result communication is paired with links that offer the PRS recipient with options to read about concepts in more detail, should they wish so.
Trust and transparency	A section is included in the design concept that gives a data privacy notice on how the recipient's genomic data is used.

Table 8: This table highlights how the user needs extracted from the literature review and interview study are applied into the digital design solution.

Genetic counseling method	**Application in the design solution**
Empathic statements	General formulation of information, used for the AI-enabled virtual genetic assistant in horizon 3 of the strategic roadmap (refer to Chapter 4.7)
Paraphrasing and summarizing (Reflection)	Chatbot function for follow-up questions and clarifications
Clarification	Opportunities for clarifying follow-up questions via the Frequently Asked Questions (FAQ) section, the chatbot, or via direct contact
Analogies	Use of a fire safety metaphor to communicate genetic risk
Giving a sense of control	The fire safety metaphor highlighting that the person can do something about the risk.

Framing genetic risk as a small change	The PRS results of My Genetic Mirror may be perceived as immediately threatening one's health and lead to an overstatement of its implications. Thus, a branding strategy is created to distance the perception of My Genetic Mirror from being directly linked to medical results with a direct threat to one's health.
Value of an early warning	The fire safety metaphor highlighting that genetic risk is about future developments, and that the PRS communication serves as a early warning.

Table 9: This table highlights how different genetic counseling methods are applied into the digital design solution.

4.6.1 Tailoring the service to support different personas throughout the user journey

An overview of the components of the service and their relevance for each persona is included in Figure 15. The following highlights how each service component relates to respective personas.

The Anxious Receiver (AR)

Although the AR prefers a consultation to discuss her PRS result, she is open to first learning about the result online, since she is used to receiving medical results online and so she can prepare questions for the consultation. For her, the most important parts are spreading out the information delivery, the brand message, the metaphor, and an actionable way to schedule a consultation after getting to know the result. The spread-out information delivery serves to not overwhelm her with information about her health. Instead, the communication aims to familiarize her with the service before the actual PRS result is announced.
After the result is announced, the digital communication aims to give her an overview of the result and provide her with an actionable button where she can schedule a consultation. The Follow-Up Questions Module (FUQM) is less relevant for her since she aims to discuss the result with a healthcare professional. However, the AR may still benefit from the FUQM, since she may get answers for her initial questions.

The Technologically Savvy

The TS values the ability to get follow-up clarification on the result via online channels. Normally, she would do internet research to understand the result. Thus, the FUQM, especially the human-powered chatbot, the AI-powered chatbot in Horizon 2, and the AI assistant in Horizon 3 are meant to support her in understanding the result via digital means. Regarding the other components, the spreading out of the information delivery, branding, and the metaphor aim to calm her down before and after the result delivery.

The Pragmatic

The Pragmatic is practical and puts a prime focus on understanding the actions she needs to take to manage her risk. She is used to being communicated medical results via digital channels. In case of high risk, she wishes for concise and reliable information for the practical steps, and if she is not provided with such, she would consider contacting a healthcare professional.

For the Pragmatic to understand the practical steps she can take to manage her risk, she would mainly benefit from the practical focus of the result delivery, lifestyle recommendations, and the FUQM. The result is formulated in a way that gives the recipient practical suggestions right away, with further lifestyle recommendations giving further practical insights, and the FUQM helping to clarify the questions that have emerged about the result and the recommendations.

The Indifferent

The Indifferent shows a minimal emotional reaction to getting a PRS result. When receiving a low PRS result, she reacts to it calmly and appreciates to-the-point explanations, but would most likely not seek follow-up information. Here, the practically-oriented results section aims to provide her with the essential information in a concise manner. When receiving a high PRS result, she places her focus on understanding the practical steps and looking for clarification. Here, like with the Pragmatic, the lifestyle recommendations and the FUQM, including the chatbot, further offer her resources to clarify follow-up clarification.

4.6.2 Service concept – elaborated

This chapter highlights the future vision based on an adjusted ViP method and gives an overview of the service and its components, including branding, metaphor usage, the FUQM, and the process of spreading out the information delivery, and shows the resulting service bluerprint (Figure 16).

In avoiding overreaction for PRS recipients, the information must not be presented in an overwhelming manner. This includes the timing of PRS result delivery. Due to low genetics knowledge among the sample (insight from the user study) and the large amount of information provided in the mock-up reports, the result can be overwhelming. Thus, in designing the service blueprint, spreading out information delivery was a key priority. When considering one's genetic risk, the person should be introduced to it gradually, and not jump immediately to highlighting all the different risk factors that a person has.

Future vision for the service

Using a simplified ViP method from the "Context and Conceptualization" course, a future vision and an elaborated vision description for the service were formulated as the following.

> **A fully digital and inclusive genetic health management service for a variety of polygenic risk**
>
> A fully digital PRS communication service that gives users a holistic understanding of their genetic health. By incorporating multiple PRS into the result, the service helps the individual and the healthcare system to identify risks earlier. An integrated AI assistant helps to give people a sense of human touch in online consultations that helps to ensure that the PRS recipients are being tended to.

"Holistic" entails the recipient being able to, after receiving multiple PRS results, get a general-level understanding of the state of one's genomic health. The future vision for the service relates to the third horizon in the tactical roadmap (Chapter 4.7). It focuses on the service offering support for PRS recipients in understanding their PRS results via an AI assistant with an audiovisual avatar. Furthermore, it relates to regular follow-up consultations to monitor the progress of the individual in managing genetic risk. Whereas the future vision in this sub-chapter relates to the service concept, the future vision in the strategic and tactical roadmaps relates to a desired state in the future (Simonse, 2018). The service concept relates to Horizon 1 of the tactical roadmap, where the roadmaps articulate the evolution of the proposed service concept throughout the horizons.

The service blueprint

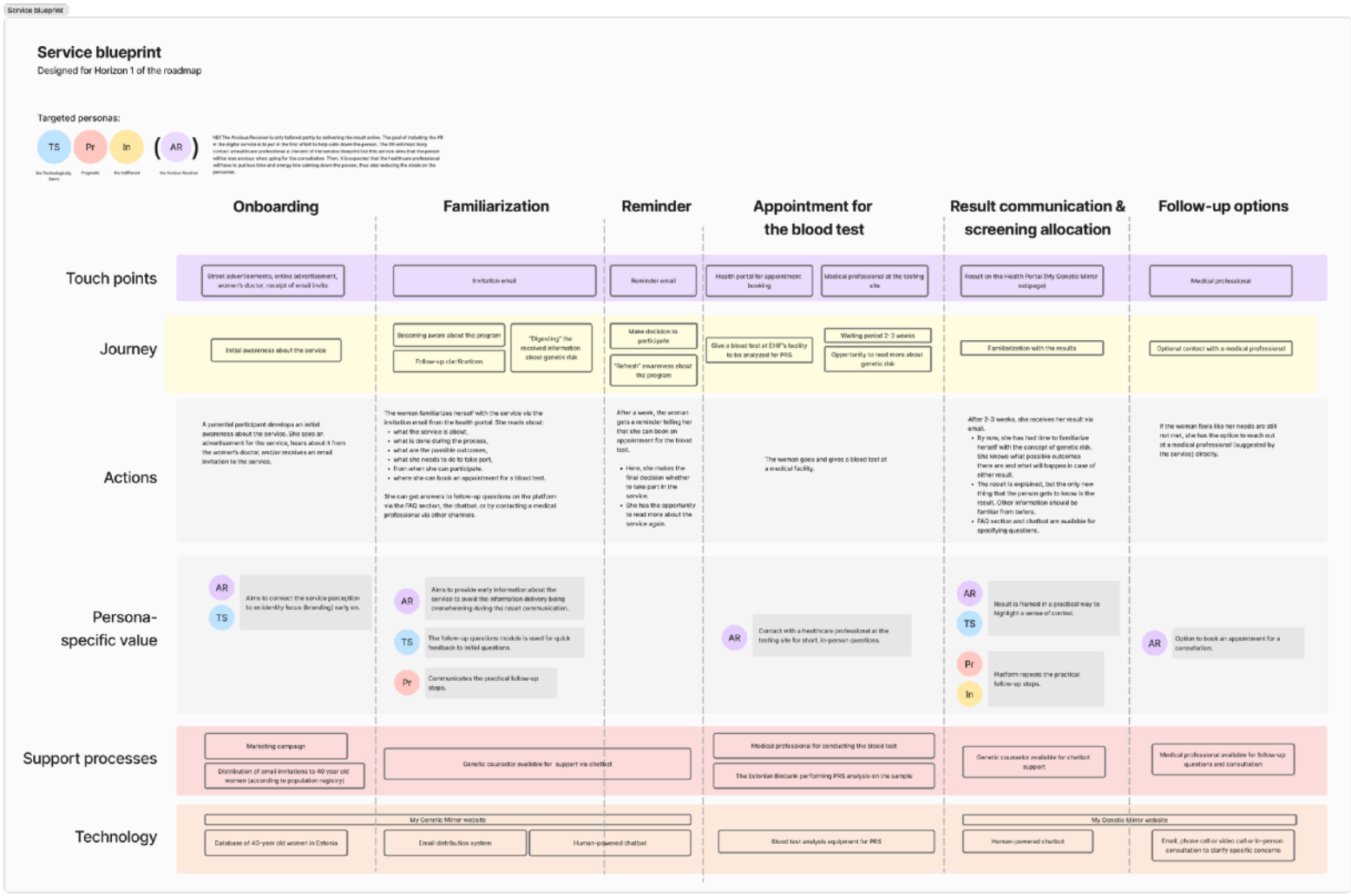


Figure 16: The resulting service blueprint.

The service blueprint (Figure 16) was based on the general structure of Bittner et al. (2008), including the touchpoints, the journey, actions, support processes, and the technology layers. To account for the tailoring to different personas, a separate layer called "Persona-specific value" was added. A service blueprint is a visual tool used to detail and analyze the service delivery process, focusing on the user's perspective and identifying key interactions between users, the service provider, and support processes. (Bittner et al., 2008)

The placement of the service in the context of the Health Portal is visualized in Figure 17. The website utilizes both email invites and a sub-page on the Health Portal ("Terviseportaal") of EHIF. This relatively conservative format for the service was chosen since it is similar to the way an invitation to breast cancer screening is organized. In designing for a wide audience, a format that users are accustomed to and that has a proven track record was chosen (Aasmaa,

2007). This digital channel is familiar to users and they are used to receiving health information via the channel, as evidenced by user interviews. The Health Portal is used by EHIF for a variety of notifications relating to health, such as appointment reminders, test results, vaccination schedules, and prescription renewals. It is supported by the national digital infrastructure, ensuring robust and reliable performance. The familiarity and reliability of the Health Portal make it an effective and trusted medium for delivering important health information, contributing to its widespread acceptance and success as a communication tool. By leveraging this well-established system, the website ensures that users receive timely and

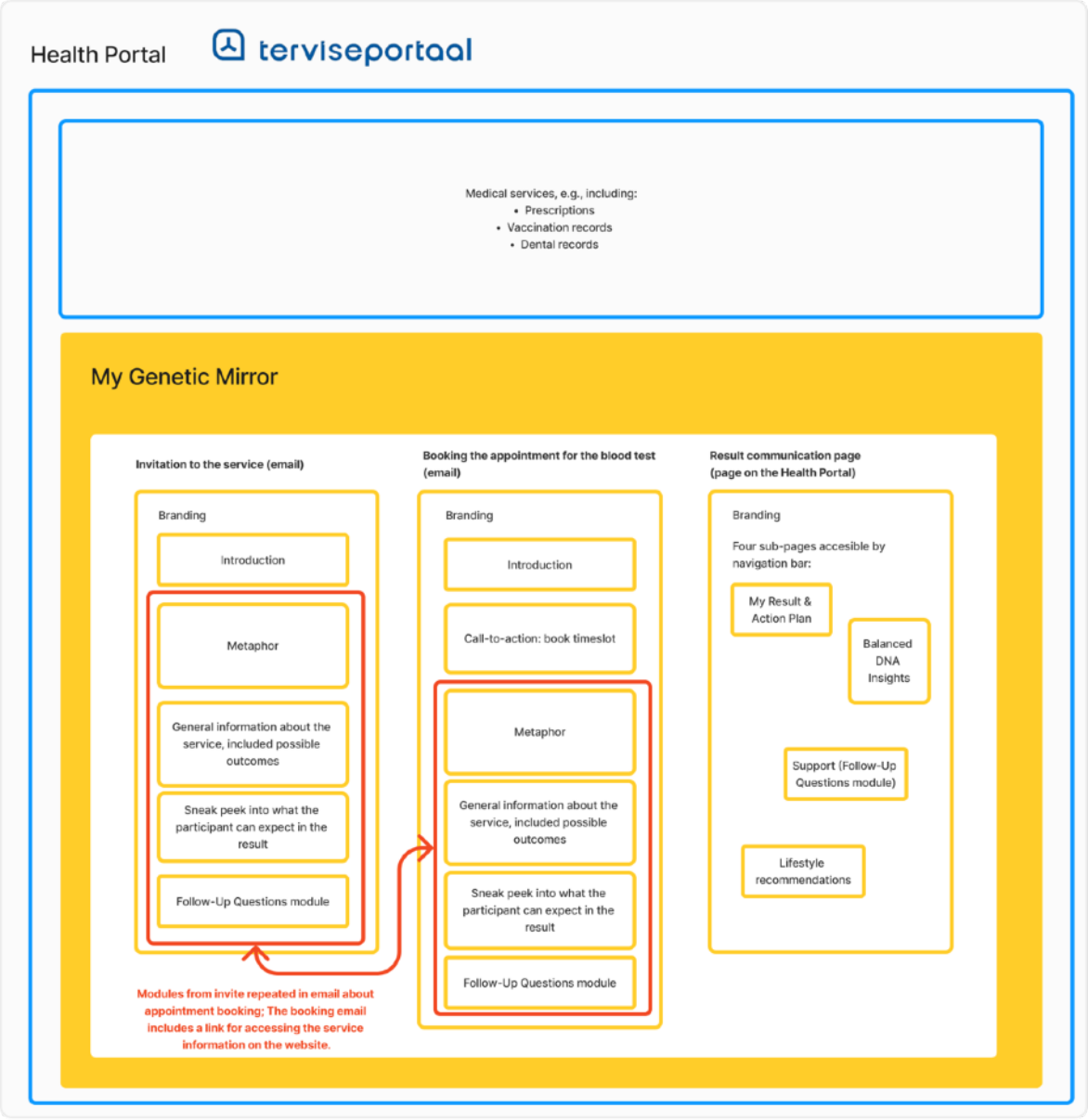


Figure 17: A schematic showing how the My Genetic Mirror service fits into the context of the Health Portal and which interfaces it consists of. The figure serves as a detailed version of the general user pathway (Figure 17b) for the stages "Email invite & familiarization", "Book appointment", and "Results delivery". Whereas the first two are delivered via email, they are also available on the My Genetic Mirror subpage on the Health Portal. The result communication page includes a navigation bar on the top of the page with buttons for four sub-pages for accessing the different parts of the result and related information. The prototype interfaces are included in the deliverables of the graduation project.

accurate information in a format they trust and regularly use, enhancing the overall effectiveness and user experience of the service.
For the website format, a partnership between the EHIF and the Health and Welfare Information Systems Centre (TEHIK; which runs the Health Portal) for the development of the My Genetic Mirror page should be started. All of the interfaces were designed according to the brand guidelines highlighted in later parts of Chapter 4.6 and the digital prototype interfaces are included in the deliverables of this graduation project.

General user pathway through the service

Figure 17b: An overview of the general user pathway through the service. This part of the service is intended for all personas.

Figure 17b shows the different parts of how the service is structured. The goal of the structure is to spread out information delivery over a longer time period so that the result would less likely be overwhelming for the recipient. The first touchpoints (advertisements, news articles, promotion when visiting a women's doctor) relate to giving a potential participant early awareness about the service. The second part relates to an email invite, where a potential participant can familiarize herself with the service, including about genetic risk. After that, a week is scheduled between the email invite and a follow-up email about booking the timeslot for a blood test (necessary for genomic analysis). After 2-3 weeks, once the result is analyzed, the participant receives the result via a notification email, which redirects the person to the Health Portal. A detailed overview schematic about the structure of the stages "Email invite & familiarization", "Book appointment", and "Results delivery" in the context of the Health Portal are shown in Figure 17.

Initial touchpoints

The aim of the initial touchpoints is to create first awareness about the service. The goal is to increase the probability that the person will engage with the service once she receives the

respective invitation in the following sub-chapter (Kronrod & Huber, 2019). Similar to the marketing strategy of other cancer screening programs in Estonia, a wide array of channels for initial touchpoints should be utilized, including advertisements (online and offline), news articles, and promotion when visiting a women's doctor. These channels emerged from the user interviews.

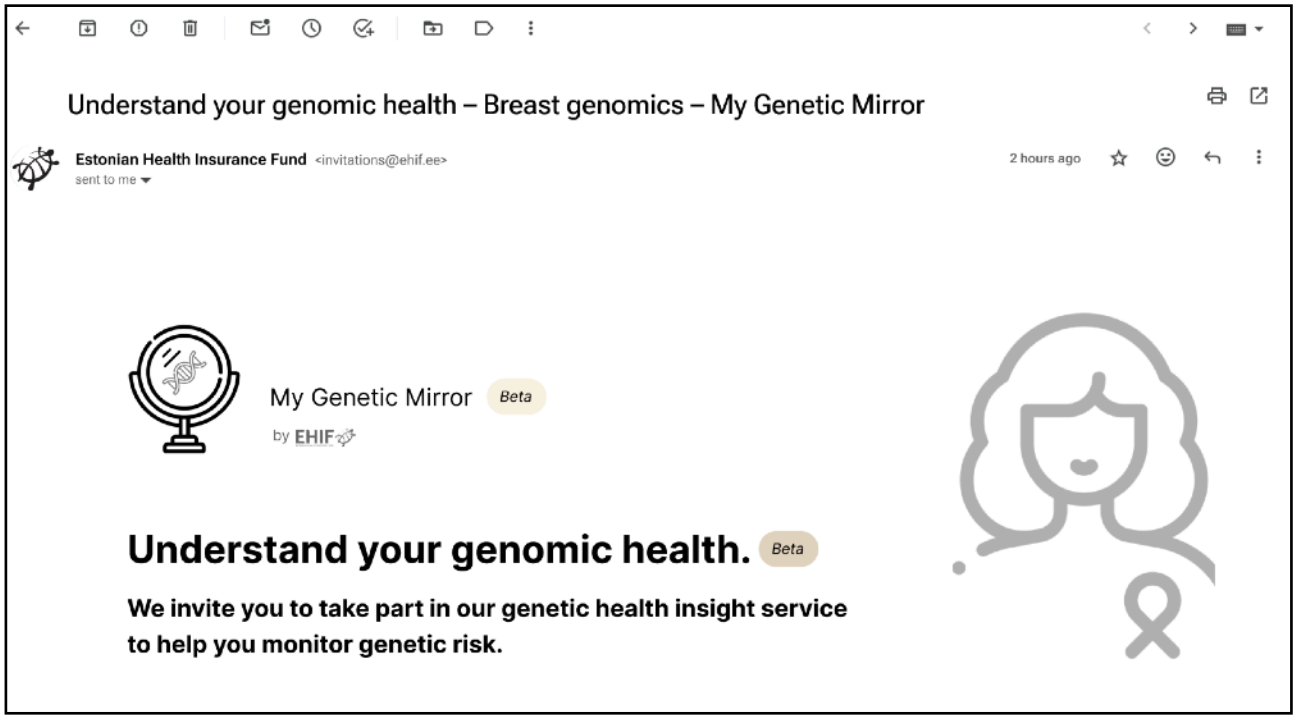


Figure 18: The beginning of the email invitation to the My Genetic Mirror service.

Email invite

The channels should be paired with an email invite to the target group to participate in the service (Figure 18). The invite serves two goals: first to invite the person to the service, and second, to increase awareness about the contents of the service. The information communicated in the invite should give the person an early but already comprehensive overview of the service. This includes the overall rationale behind the service and its contents, why the email recipient is invited, what she needs to do to participate when she can participate, and importantly, what are the potential outcomes. This is paired with the use of a metaphor to help understand (create a mental model) the meaning and the implications of the risk. Here, a fire safety metaphor aims to communicate a complex result using a relatively simple metaphor (refer to chapter 4.6.4). Here, a mirror is explained to "reflect" the recipient sitting in the living room at home. The accompanying text of the metaphor highlights that the service can be thought of as predicting the chance of there being a fire in the person's home. If a high PRS result ends up being the case for the person (a prediction), then screening can be

considered as a monitoring precaution to reduce the risk (like a smoke detector in the case of fire). The breast cancer screening program is not part of the service, but it is included in the metaphor since it relates to follow-up steps. The goal of accompanying the invite with a

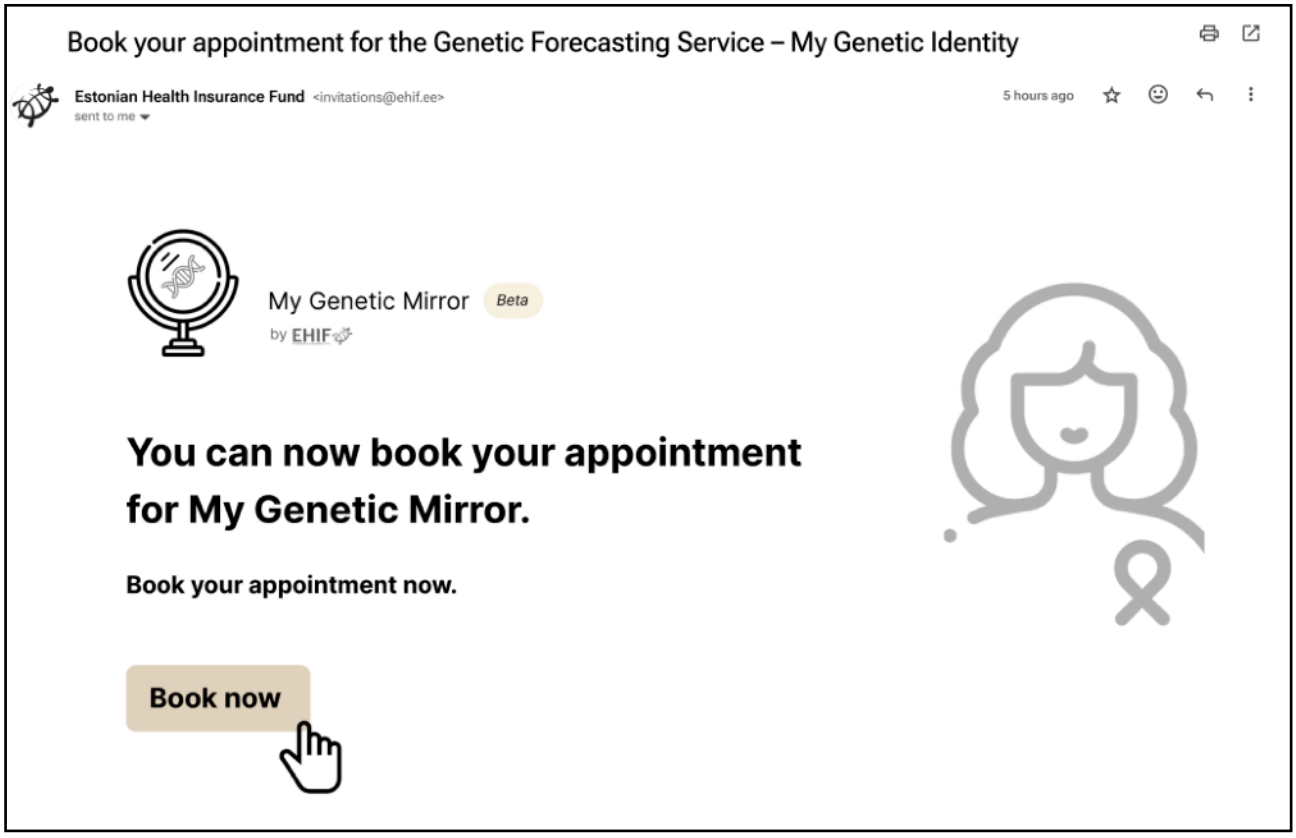


Figure 19: Screenshot from the appointment booking page for the blood test.

metaphor aims to enable the person to be able to understand the concept of genetic risk and potential outcomes before the actual result communication, helping to avoid overreaction due to an overwhelming medical information delivery.
The recipient is also provided with lifestyle recommendations and insights about the genome (which is currently left blank because for this determining the result is necessary but serves as a sneak peek into what kind of information the recipient can expect post-result).

Booking the appointment for the blood test

The invite announces the date from which the person can participate in the service (Figure 19). Once the date arrives, the person can schedule the appointment for the blood test. It is further highlighted that there should be no rush to the service since the results do not directly affect one's health. The rationale for sending out the invite before the person can participate is to allow some space for the person to "digest" the information and possibly ask follow-up questions.

Once the date for participation has arrived, the person is provided with a reminder email, asking to schedule the appointment via the online registry. Once the person has given the blood test at the appointment, there is an expected 2-3 weeks until the result is announced (the time frame is shorter for those who have allowed to use their genomic data from the Biobank).

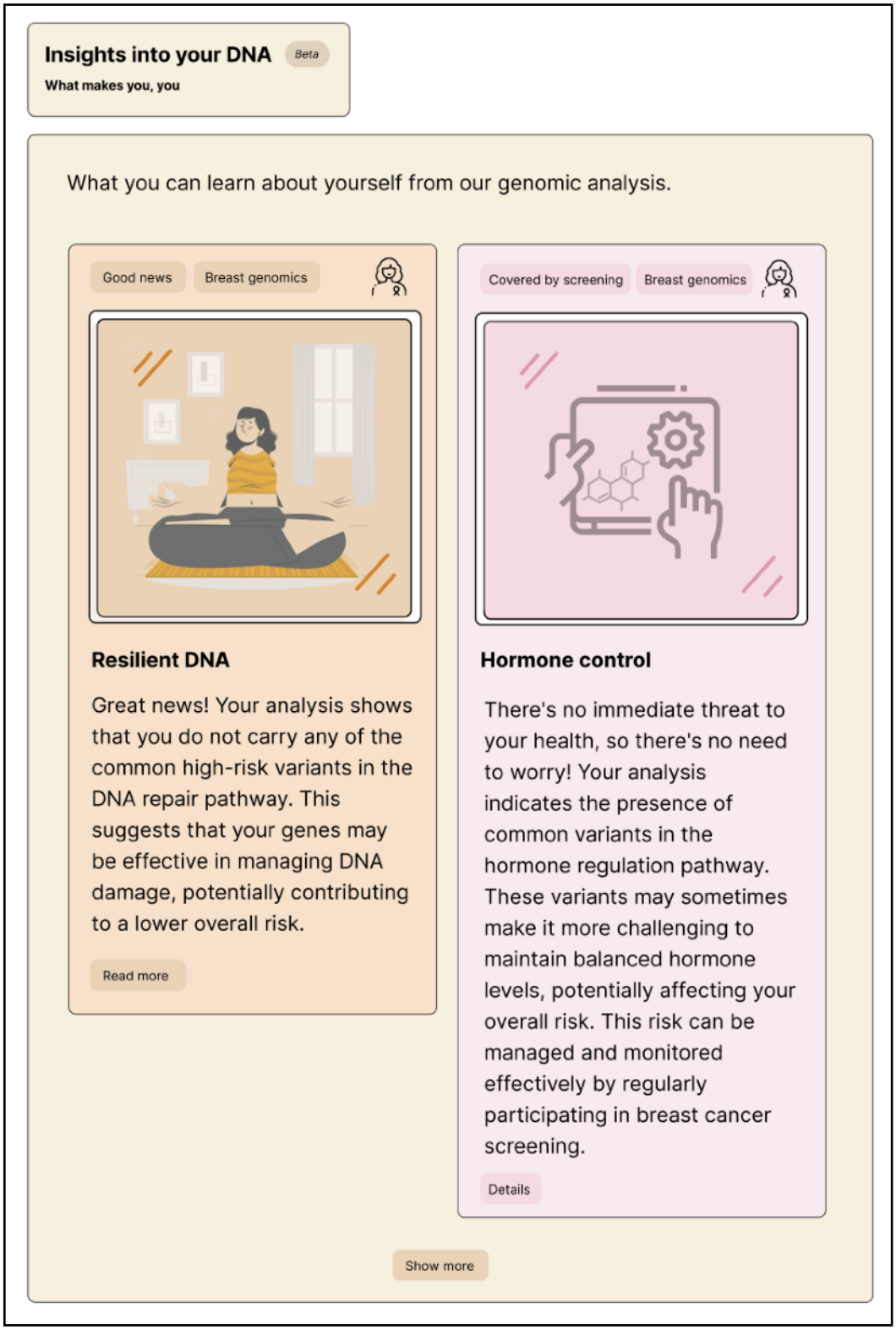


Figure 21: Part of the PRS result communication, highlighting both positive and negative insights into a person's DNA. This is connected to the brand concept, which frames genetic risk consisting of both positive and negative aspects. It furthermore enables further insights to the result, making it especially relevant for the Technologically Savvy persona.

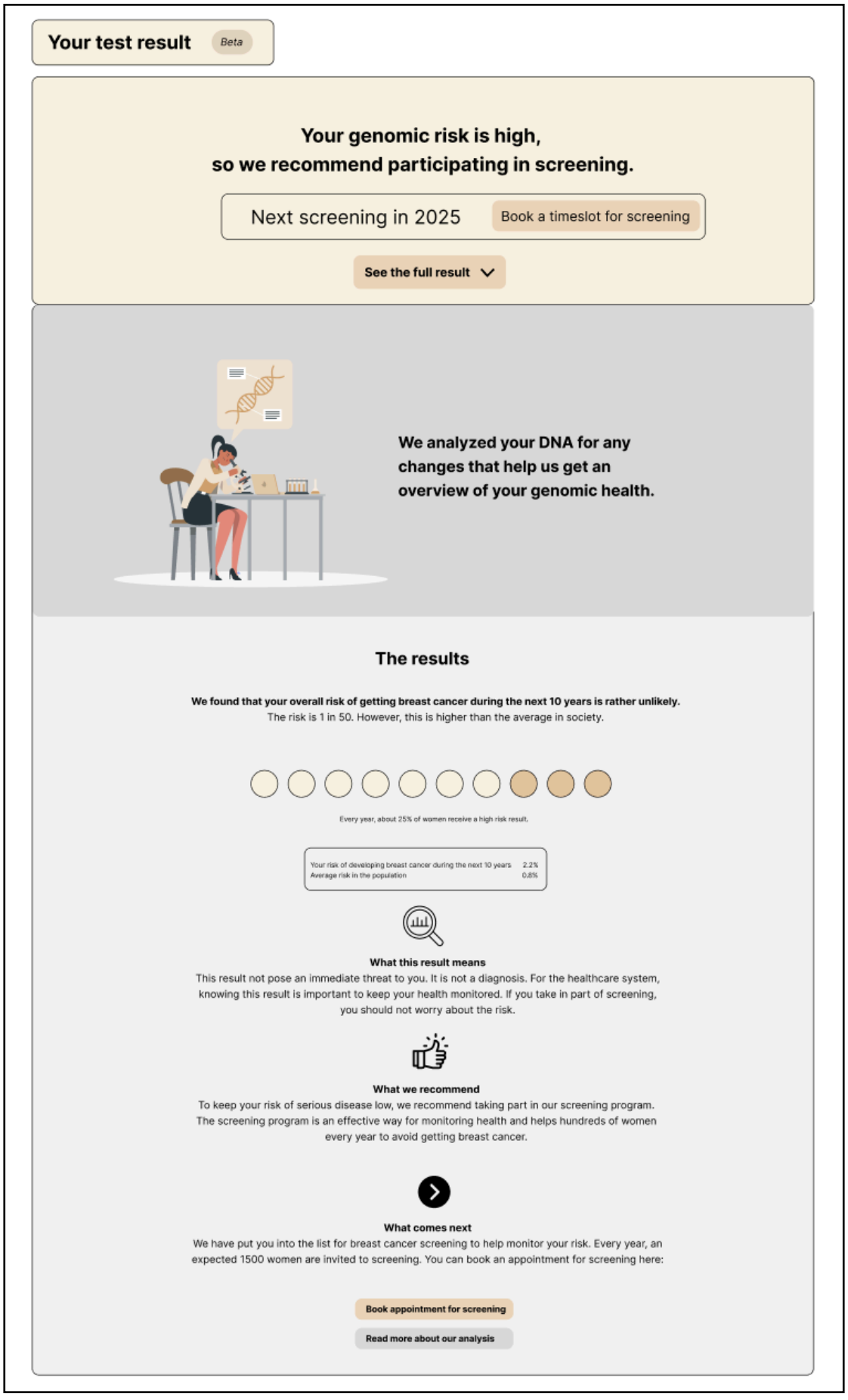


Figure 20: Overview of the results section.

Result communication

The result is communicated via the Health Portal, for which an email notification is also provided. The email notification includes a link where the recipient can access the report (Figure 20) on the Health Portal. In the result communication part, the information provided is similar to the communication in the invitation but with an added result section. First, a disclaimer is mentioned to ensure the person not mistaking genomic risk for a diagnosis, and that aims to pre-emptively calm the person down. Secondly, an interpretation is provided that summarizes the result in a short and positive manner. This includes the notification about whether a screening is intended for the person or not, and if yes, a reminder set for participating in a screening program.

The person has the option to look into the detailed medical report by clicking on the dropdown "See the full report". The medical report should respond to the information needs of the recipients. However, since the final medical report should be made by genetic experts, the following highlights guidelines from the user study for the formulation of the report:

- The information in the report should be clear and straightforward,
- medical jargon and complex statistical explanations should be avoided,
- include a simplified summary and a visual aid, and
- provide a conclusion on recommended follow-up guidance, and
- although the methodology and model limitations should be used, they should be put at the end.

In the medical report, the user can read into detail about the result, for which the mock-up only provides a simplified example formulation. The reason for the dropdown is to ensure that the person first reads the short result summary above before looking into the exact report. This was done to avoid misinterpretation of the medical result, since by first seeing the detailed report, the recipient may not understand it and overreact. The decision was based on an insight from Interviewee 14 (expert), who recommended focusing on follow-up action (assignment to screening) since recipients may interpret a medical report differently. Furthermore, as interview respondents expressed a wish to learn which parts of their genome contribute to the risk, an overview of both positive and negative insights is provided (Figure 21) These information pieces are meant to be extracted from the PRS analysis. As an example, if the results show that the recipient does not have a mutation in the SNPs related to BC PRS (Wang et al., 2018), then the recipient can consider this positive. This can highlight to the recipient that there is also a reason not to worry since there is positive news included as well. The rationale behind including both positive and negative information lies in providing recipients with a balanced overview of their genetic health. Self-concept clarity (SCC; Campbell et al., 1996) refers to the extent to which the individual's self-beliefs are stable over

time. If the recipient's result indicates a high PRS, this, on its own, can be considered negative news, possibly impacting the individual's perceived self-belief about her health and leading to lowered SCC. An empirical study by Jiang et al. (2023) suggests that low self-concept clarity inhibits self-control. In turn, low self-control is associated with frequent rushing and hurrying (Baumeister et al., 2018), which may also affect the PRS recipient's likelihood of reaching out to a medical professional.

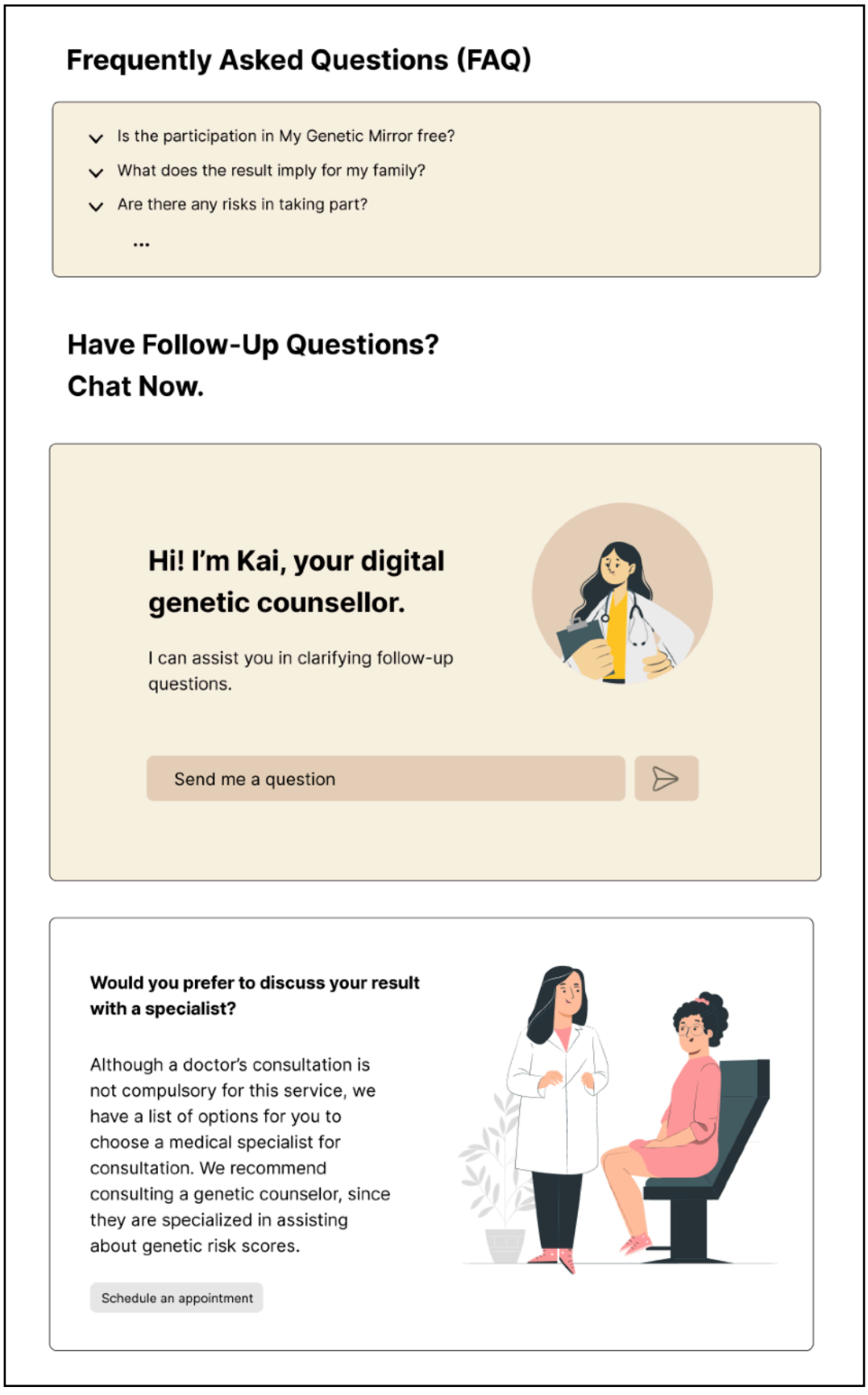


Figure 22: The Follow-Up Questions module showing FAQs, a chatbot, and additional contacts for booking a consultation with a healthcare professional.

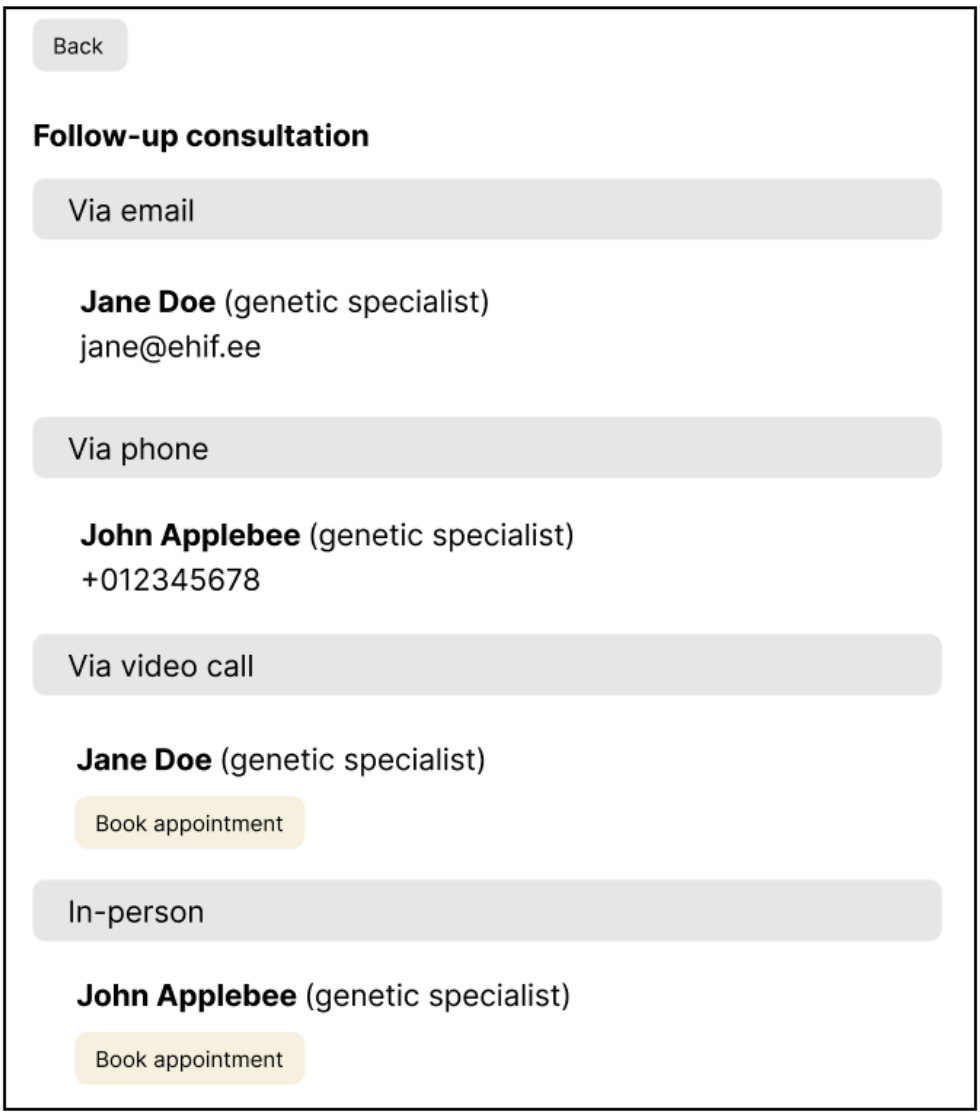


Figure 23: The pop-up contacts section to a medical professional (left), when clicking on link "see contacts" (see Figure 22).

The Follow-Up Questions Module

The follow-up questions module is meant to be used both before and after the result communication. Before the result communication, the module aims to clarify questions about participating in the service as well as follow-up questions about genomic risk. After the result communication, the module is also given in order to clarify additional questions that the person may have after the result communication. The module is shown in Figure 22.

The module consists of three parts: a Frequently Asked Questions (FAQ) section, along with the option to ask specific questions via a chatbot, and a list with further contacts. User interviews showed that although some respondents may prefer an in-person consultation, others mentioned that having a quick interaction with a specialist to clarify specific questions to be beneficial. In case of these specific questions, the chatbot module is provided, along with email contacts. The chatbot should be operated by a genetic counselor who has the qualifications to answer questions about genetic risk. Although there is currently only one genetic counselor in Estonia, expert interviews showed that there are multiple genetic counselors in training. Experience via chatbot interactions with users can constitute a valuable source for learning about how to communicate genetic risk as well as to contribute the findings to academic literature.

For interacting with the user via the chatbot, the genetic counselor can utilize methods that (s)he normally uses in consultations. According to findings from the literature review and the user study, these can constitute empathic interaction, framing PRS as a small change for the person, emphasizing a sense of control, highlighting behavior change opportunities, and the value of getting an early warning. The chatbot is intended for more general questions. Thus, if the text-based format does not suffice for consulting the user sufficiently, the genetic counselor can propose an alternative communication channel, e.g., a video call.

If the information provided so far does not suffice for the recipient, then recommended contacts to genetic specialists intended to assist as part of the service are provided. It is highlighted to the recipient that a doctor's consultation is not compulsory for this service but if there is a specific and urgent question nonetheless, the contacts can be used. The contacts are ranked top to bottom: contact via email, phone, video call, and only then in-person consultation (Figure 23). This contact section serves two purposes: first, to tell the recipient that these contacts are intended only when (really) necessary, and to provide further options in addition to in-person consultations. The rationale here is that a person may get their question(s) answered via a more cost-efficient channel, e.g., an email instead of an in-person consultation, and in turn, reduce the strain on the healthcare system.

Figure 24: The fire safety analogy in the results section, along with the mirror giving a "reflection" of the person's home, which the metaphor parallels with the human body.

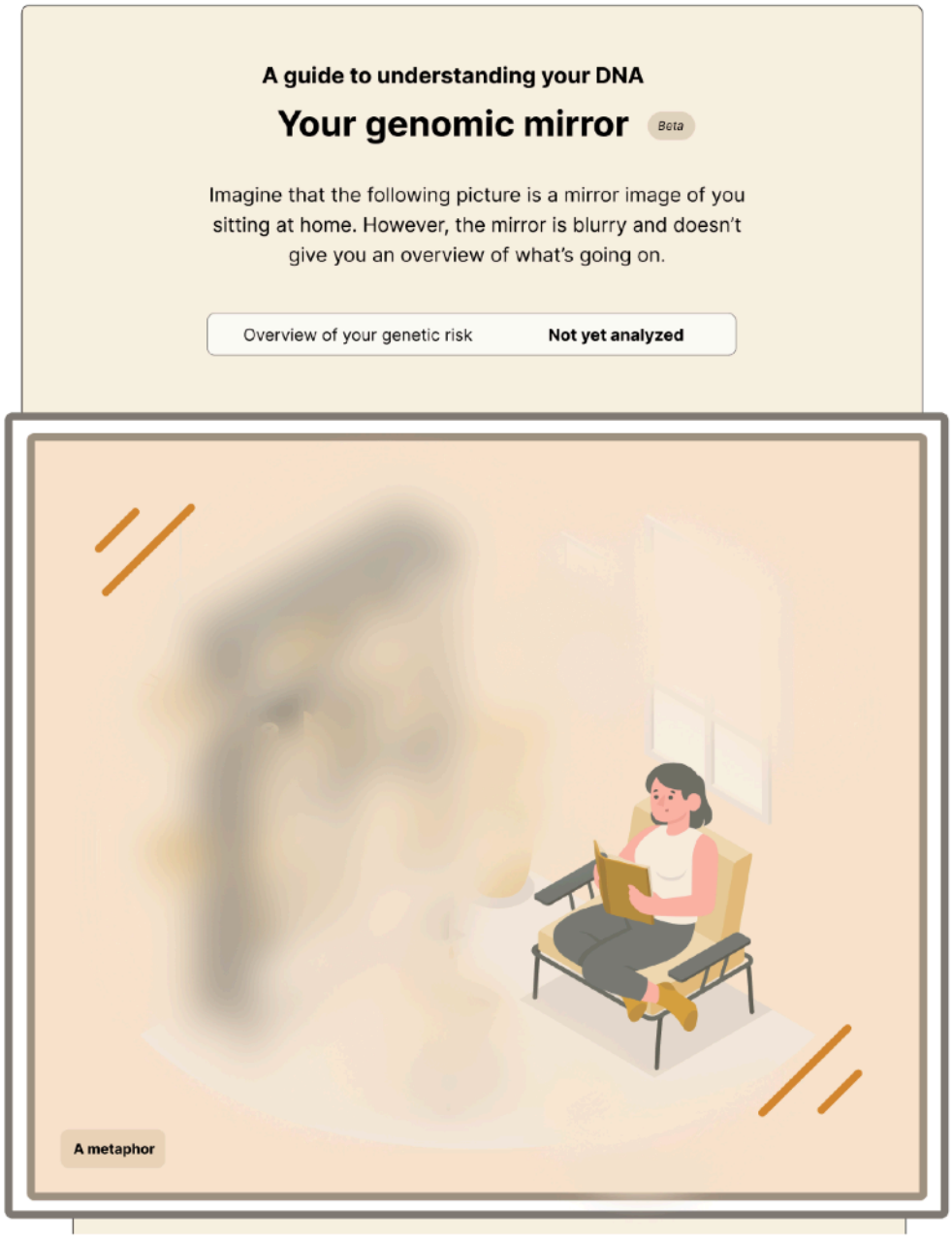


Figure 24b: The fire safety analogy in the invitation email, showing a mostly blurred mirror. The above text box highlights that the genetic risk is not yet analyzed and thus the mirror cannot provide an overview. The rationale is to motivate a person to take part in the service to find out the full mirror image.

4.6.3 Communicating genetic risk via a metaphor

In this chapter, the fire safety metaphor used to communicate the service is described (Figure 24). In this sub-chapter, the rationale for using a metaphor is first explained, after which the process of coming up with the metaphor is elaborated. According to Kövecses (2010), metaphors are not only shaped by but also shape cultural perceptions. Landau et al. (2018) highlight that metaphors in health messages can shape cognitive processes and influence perception and behavior. They argue that metaphors provide a structured way of understanding complex and abstract health-related information by mapping it onto familiar concepts. For example, using the metaphor of "fighting a battle" to describe cancer can help patients conceptualize their treatment as an active struggle. Sexton & James (2021) highlight that metaphors simplify complex genetic concepts into personally meaningful terms, enhancing understanding of clients' experiences and emotions. They also aid in processing these emotions and concepts, helping both client and counselor explore subconscious feelings, self-concept, and motivations. For instance, they describe metaphors frequently heard in genetic counseling, such as "a ticking time bomb" to describe genetic predisposition, "a rollercoaster" to describe the genetic diagnosis process, and 'a weight has been lifted' to describe decision-making moments. These metaphors help clients to better understand and emotionally process their genetic information.

Martin et al. (2023) emphasize the importance of using metaphors to compare uncertain and unfamiliar experiences with familiar references, specifically in communicating genetic risk. This approach can aid individuals in challenging health-related situations by facilitating sense-making and helping them communicate complex emotions. They present multiple metaphors in which women perceive genetic risk upon communication, including "knowledge is power," which empowers individuals by providing them with information they can use to make informed decisions about their health; the "gambling" metaphor, which conveys the uncertainty and risk involved; and the "journey" metaphor, which emphasizes the ongoing and evolving nature of living with genetic risk. Additional metaphors presented by Martin et al. (2023) include "rollercoaster," "battle, disaster, or wreckage," "Pandora’s box or a can of worms," "doom and gloom," and "the release or placing of a weight."

Each of these metaphors captures different aspects of the emotional and cognitive experiences of individuals undergoing BRCA gene testing. The "rollercoaster" metaphor describes the tumultuous and emotional ups and downs experienced during the testing process, highlighting the varied and intense feelings that can arise at different stages. The "battle, disaster, or wreckage" metaphors convey the sense of conflict, chaos, and destruction that can accompany receiving a BRCA-positive result, emphasizing the disruptive impact on an individual’s life and sense of security. The "Pandora’s box or a can of worms" metaphors illustrate the fear of uncovering potentially overwhelming and uncontrollable information,

reflecting the anxiety associated with genetic testing and its implications. The "doom and gloom" metaphor captures the feelings of despair and hopelessness that can result from a BRCA-positive diagnosis, emphasizing the emotional weight of the information. The "release or placing of a weight" metaphor describes the relief or burden experienced when receiving test results, highlighting the emotional significance and the impact on an individual's mental state.

Figures 25 & 26: The participants at the creative facilitation session.

Spiegelhalter (2016) suggests the effective age metaphor, which entails comparing a given individual's health with that of a 'healthy' person with the same risk profile. This relates to a finding from an expert interview, where he stated that genetic risk can be communicated similarly to the effective age metaphor. There, e.g., if a 40-year-old woman has a higher genetic risk for breast cancer, that can be communicated as if the risk of the woman is similar to that of a 50-year-old woman.

Creative facilitation session for metaphor exploration

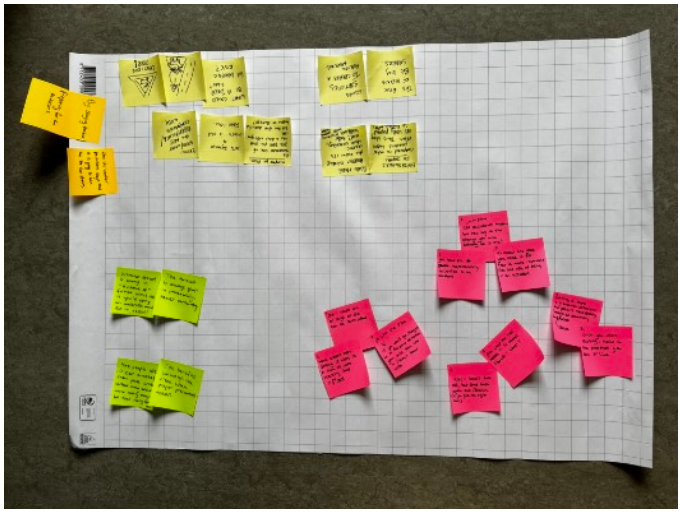
To explore further metaphors that can be applied in addressing the problem statement, a creative facilitation session was organized. Himaki et al. (2024) note that analyzing user metaphors can greatly enhance the idea-generation process by providing insights into user perceptions and behaviors, thereby improving user experiences in various ways. The goal of the creative facilitation (CF) session was to explore cognitive constructs, specifically metaphors, and analogies, which the recipient of a PRS result can use to make sense of their health situation. For preparing for the CF session, the book "Road Map for Creative Problem Solving Techniques" (Heijne & van der Meer, 2019) was used. Consent for participation was obtained by using the result Post-It notes and audio recording in the analysis and conceptualization of the thesis. The CF session was performed with four participants, all with

a design student background (Figures 25 & 26). The session focused on the idea-finding stage of creative facilitation, with the Creative Confrontation method with the Direct Analogy or Metaphor excursion used. The goal of the method was to start with brainstorming more obvious ideas, with the excursion guiding the participants to explore the problem setting on deeper layers of knowledge and to generate options beyond the obvious using analogies and metaphors.

The problem statement for the session was formulated as "How may we frame genetic risk in our heads in the context of everyday life?". The various metaphors and analogies generated during the session (Figures 27 & 28) include natural phenomena, weather forecasting, accidents, and natural disasters. The metaphors about natural phenomena focus on how typically negatively perceived phenomena like storms on a sea can happen, but that doesn't mean that they will last forever. Moreover, a person's health was likened to a plant, that should be given the needs to flourish. If a plant, e.g., a tree, is not looked after, its leaves will fall off; However, if the plant is watered, its leaves will grow back.
The metaphors about forecasting focus on how the forecast is always provided in probability and never in certainty. According to the participant, the process of monitoring one's health in case of genetic risk can be compared to bringing an umbrella when rain is expected. Thus, in the worst case, one would be carrying around an umbrella for no reason.
The metaphors about accidents were related to plane and shark attacks. These emphasize that a person may overstate the risk level of certain accidents, e.g., plane crashes and attacks by sharks. The metaphors about natural disasters brought earthquakes in Japan as an example. Since Japan is more prone to earthquakes than other countries, the country is also more prepared. Moreover, if one is driving a car in a dangerous region, there can often be a road sign suggesting caution when navigating. It was thus suggested that such a "caution warning sign" equivalent could be there for genetic risk. For example, if a person gets to know that have a higher risk, such a "warning sign" of a dangerous region could metaphorically be presented to them. A similar example was brought with the Netherlands using dams to prepare for floods and storms. Overall, a person with a higher risk of genetic risk should be more prepared.

Aligning the metaphor with user needs

To select the suitable metaphor(s) for digital communication, it should align with the needs of users in different stages of the service. For this, the mapped user journey from the introduction is referred to.
During the first stage, a potential participant has received an invite to participate in the program but hasn't decided to participate yet. Here, it is crucial to explain to the person the

Figures 27 & 28: The results of the creative facilitation as Post-It notes.

essence of the program, which lies in PRS testing. To explain the concept with a metaphor, the latter should communicate...

- that there are (genetic) variables outside the control of the person,
- the result can be both positive or negative,
- that the PRS result is stochastic,
- the recipient can understand the metaphor from their perspective, and
- a metaphor that is widely recognized by the general population.

After receiving feedback from a supervisor, an additional criterion was added: the metaphor should be on a micro-level, i.e. that it should be relatable to a person on a small-scale, human body context. Thus, another round of individual brainstorming for further metaphors was done, resulting in the following metaphor being chosen: genetic risk, the PRS communication service, and follow-up monitoring altogether can be understood as a smoke detector in case of a fire at a person's home. This metaphor was also chosen since it relates to an ongoing and well-known home fire safety campaign (since 2018) in Estonia called "Making Homes Fire-Safe" ("Kodud tuleohutuks") by the Estonian Rescue Board (Päästeamet, n.d.; Projekt „Kodud Tuleohutuks", n.d.; Figure 29). The campaign highlights the importance of fire safety precautions at home, including maintaining smoke detectors. Thus, the importance of having a smoke detector and fire safety precautions at home should not be unfamiliar to the wider Estonian public.

Figure 29: An advertisement for the "Making Homes Fire-Safe" campaign (Kodud tuleohutuks, 2024). The campaign, along with the visual, serves to emphasize the importance of taking fire risk more seriously.

The metaphor conveys that the risk of having a fire at home is low (just like getting breast cancer even after a high PRS result, which is only about 2%) and that the smoke detector (the PRS service and subsequent BC screening) notifies the person if there actually is a fire developing (breast cancer). Here, the PRS communication service is conveyed as a future prediction of having a fire in one's home. The fire safety metaphor further highlights that it is possible to avoid a fire if the protection against fire is regularly maintained, e.g., by replacing batteries in a smoke detector. It thus highlights the main ways in which genetic counselors explain genetic risk: PRS as a small change (low chance of fire), the value of an early warning (already getting the PRS result about future conditions), sense of control and behavior change (taking part in monitoring).

Goal of the fire safety metaphor

The goal of this metaphor is not to evoke fear but to inspire a proactive attitude toward genetic risk management. This metaphor is chosen to convey two main messages: the importance of taking genetic risks seriously and the reassurance that with proper precautions, these risks can be effectively managed.

By recognizing the potential danger of a house fire, individuals should acknowledge the significance of genetic risks. Ignoring these risks can lead to severe consequences, much like neglecting fire safety measures in a home. This comparison highlights the need for awareness and proactive measures in both scenarios.

The metaphor also highlights that, similar to fire risk, genetic risk can be minimized through appropriate precautions. For homeowners, installing smoke detectors, maintaining fire extinguishers, and having an emergency plan are vital steps to reduce the likelihood and impact of a fire. Similarly, individuals can manage their genetic risks by undergoing regular

screenings, making informed lifestyle choices, and following medical advice. This parallel emphasizes that while the risk exists, it is manageable if addressed with proper care.
By presenting genetic risk alongside the more familiar concept of home fire risk, the metaphor aims to make the idea of genetic risk management more relatable and actionable. It bridges the concept of genetic risk with concrete actions that people already understand and practice in other areas of their lives. This approach aims to encourage individuals to take practical steps towards managing their health risks, thereby reducing potential threats. By understanding that both genetic and fire risks can be mitigated through preventive measures, individuals can feel empowered to take control of their health, much like they do with their home safety.

4.6.4 Perception change

Problem with the service name and perception

Currently, the program's proposed name is the "personalized breast cancer screening program". However, conducted interviews indicate that the name may lead users to connect the service with aspects that are not present. For example, they may perceive it as the actual breast cancer screening program, where mammography is performed. Moreover, the person may not understand inherently that the outcome of the service is not a diagnosis. If the person overestimates the impact of the service on their health, there can be a higher chance for overreaction. In turn, the name of the service is a key factor leading women to reach out to a medical professional for a consultation.

However, the EHIF aims the service to help make genetic insights for health management available to a wider audience, while at the same time keeping it cost-efficient. This includes the rationale of performing PRS delivery online with the goal of clarifying the situation without organizing cost-inefficient in-person consultations. Thus, the perception of the service may lead people to overstate its implications on the person's health, which is a significant problem.

To change the perception of a product or service in the minds of customers and other stakeholders, branding is a commonly used tool (Mark & Pearson, 2001). Branding establishes a distinctive identity for a product (or here: service), helping to form a connection with it (Basu & Wang, 2009). To achieve this, first, a 4C (company, context, competitors, customers) analysis was conducted that focuses on EHIF as well as a more specific analysis of the Health Portal (Terviseportaal), in order to analyze the two entities before the respective design part. The 4C analysis is based on the material from the IDE course "Brand and Product Commercialization".

4C analysis of EHIF

Company (Institution)

The Estonian Health Insurance Fund (EHIF), known as Tervisekassa in Estonian, is the central organization responsible for administering health insurance and managing healthcare funding in Estonia. Established in 1992, EHIF operates under the Ministry of Social Affairs and is tasked with ensuring that all insured individuals receive necessary medical care. EHIF's mission is to provide access to quality healthcare services, promote health, and ensure the efficient use of healthcare resources. The organization oversees the allocation of financial resources to healthcare providers, negotiating contracts, and ensuring the provision of services according to set standards. The EHIF covers approximately 95% of Estonia's population, funding health services through social health insurance (SHI) contributions, which constitute about two-thirds of its funding, with the remainder coming from direct state

budget transfers. Balancing the distribution of limited resources while ensuring quality and access remains a constant challenge due to increasing healthcare costs and an aging population. Leveraging digital solutions and e-health services, EHIF has streamlined processes, reducing administrative burdens and improving service delivery. (Kasekamp et al., 2023; OECD, 2023)

During the last few years, the Estonian Health Insurance Fund (EHIF) was rebranded from Haigekassa (Est. "sick" + "[insurance] fund") to Tervisekassa (Est. "health" + "[insurance fund]"). This strategic rebranding was done to better reflect the institution's mission of disease prevention and health promotion. Before, the service could have been perceived as "an [insurance] fund for the sick", but after the rebranding "an [insurance] fund for 'receiving' health". The new name, reflecting a focus on health results, was considered during the health insurance system's establishment in the early 1990s but only adopted recently to better represent the fund's mission and activities. Rain Laane, the head of Tervisekassa, explained that the name change aims to highlight the fund's proactive role in health outcomes rather than merely paying for treatments. The overarching goal is to help people stay healthy longer and reduce the need for medical attention by fostering personal responsibility for health and ensuring high-quality, timely medical care for all. Thus, EHIF has been framing its brand message in a more positive outlook on health and prevention. (EHIF, 2023).

The Estonian Health Insurance Fund (EHIF) engages in several strategic partnerships to enhance its service delivery, particularly in the field of genomics and personalized medicine. These collaborations leverage advanced technologies and research to improve healthcare outcomes for the Estonian population. EHIF collaborates closely with the University of Tartu and the Estonian Biobank. This partnership facilitates the integration of genomic data into public health initiatives. The Estonian Biobank, which contains genetic data from over 200,000 individuals, is pivotal in supporting research and personalized medicine projects. EHIF uses this data to help identify genetic risk factors for various diseases, enabling more tailored and effective healthcare solutions (Paraskevopoulos, 2022).

Antegenes, an Estonian health-tech startup, is a significant partner in advancing genomic medicine. EHIF supports the integration of Antegenes' genetic testing services into the national healthcare system. Antegenes utilizes polygenic risk scores to assess individuals' susceptibility to cancers such as breast, prostate, colon, and melanoma. This collaboration aims to enhance cancer prevention and early detection efforts in Estonia. Antegenes has received substantial funding and grants, including from EIT Health and the Norway Grants Green ICT program, to expand its operations and research in personalized cancer prevention (Allen, 2022; Antegenes, 2023).

Context

Estonia's healthcare system is centralized, with healthcare providers operating under private law but heavily regulated by the state. The system is primarily funded through a payroll tax, and public health expenditure has been increasing steadily. Despite this, Estonia's health spending remains relatively low compared to the EU average, both in terms of per capita expenditure and as a share of GDP. Recent reforms have focused on broadening the health insurance revenue base, strengthening primary care, and improving mental health services. (Kasekamp et al., 2023; Kasekamp et al., 2024)

Customers

The primary customers of EHIF are the residents of Estonia, who benefit from the health insurance coverage provided. This includes a broad set of benefits such as general healthcare services, preventive measures, and specific treatments. The fund ensures that insured individuals have access to necessary medical services without significant out-of-pocket expenses, although these expenses remain relatively high, particularly for dental care and outpatient pharmaceuticals. (Kasekamp et al., 2023; Kasekamp et al., 2024; EHIF, 2024). The fund's customer base is extensive, covering nearly the entire population, which makes it a critical component of the Estonian healthcare system (EHIF, 2024). The conducted user interviews highlight that the respondents generally trust the Estonian healthcare system, in which EHIF is a central player.

Competition

While EHIF is the predominant player in Estonia's public health insurance sector, it faces indirect competition from private healthcare providers and insurance companies offering additional or faster services for those who can afford them. One notable competitor in the realm of personalized medicine is Antegenes, an Estonian health-tech company specializing in genetic testing for cancer prevention. Antegenes uses innovative polygenic risk score technology to assess individuals' genetic predispositions to cancers such as breast, prostate, colon, and melanoma. This personalized approach offers more accurate prevention and early detection measures, posing a modern alternative to traditional healthcare services provided by EHIF (Allen, 2022; Antegenes, 2023).

Antegenes' services are integrated with Estonia's healthcare infrastructure, collaborating with the Estonian Biobank and other medical institutions to provide comprehensive genetic risk assessments. This collaboration ensures that their services are accessible and beneficial to a

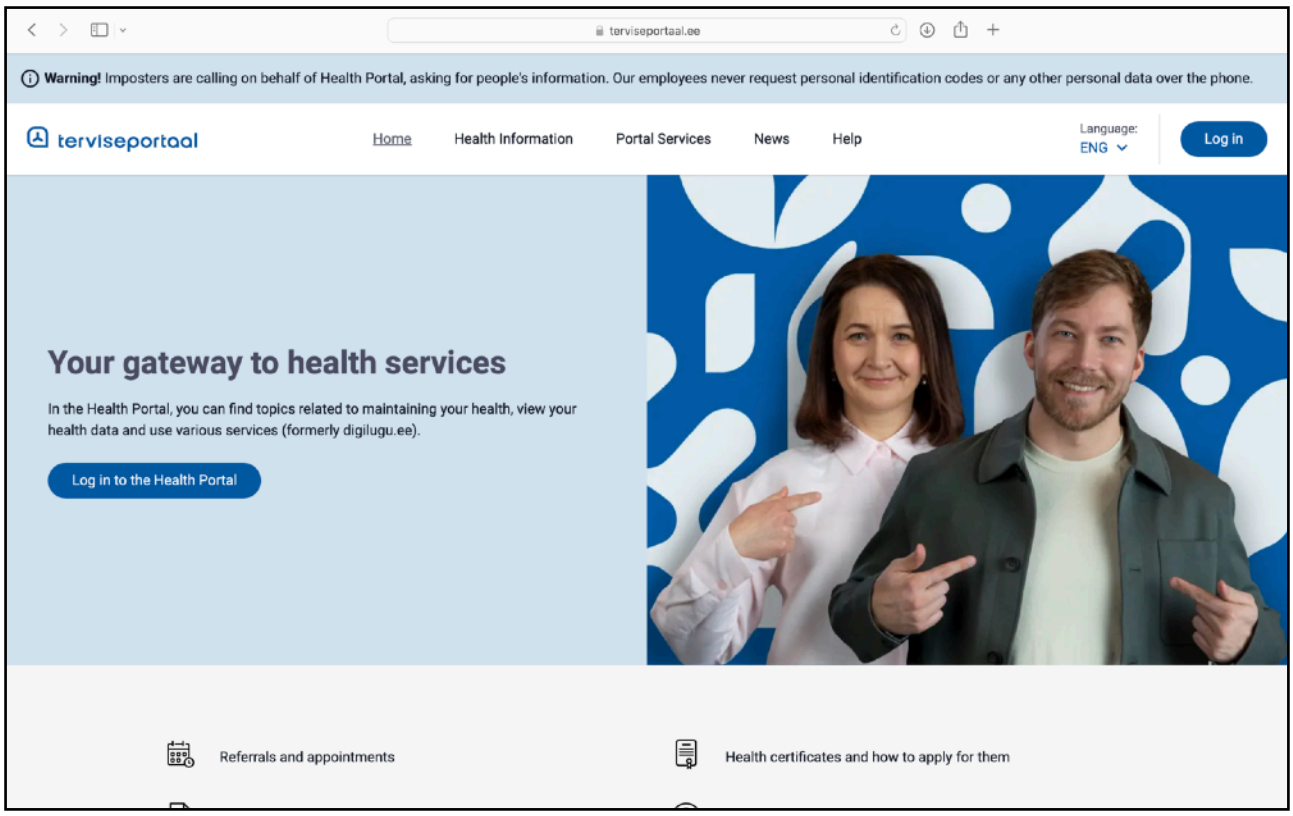


Figure 30: The landing page of the Health Portal. Sensitive information has been removed from the screenshot due to privacy concerns.

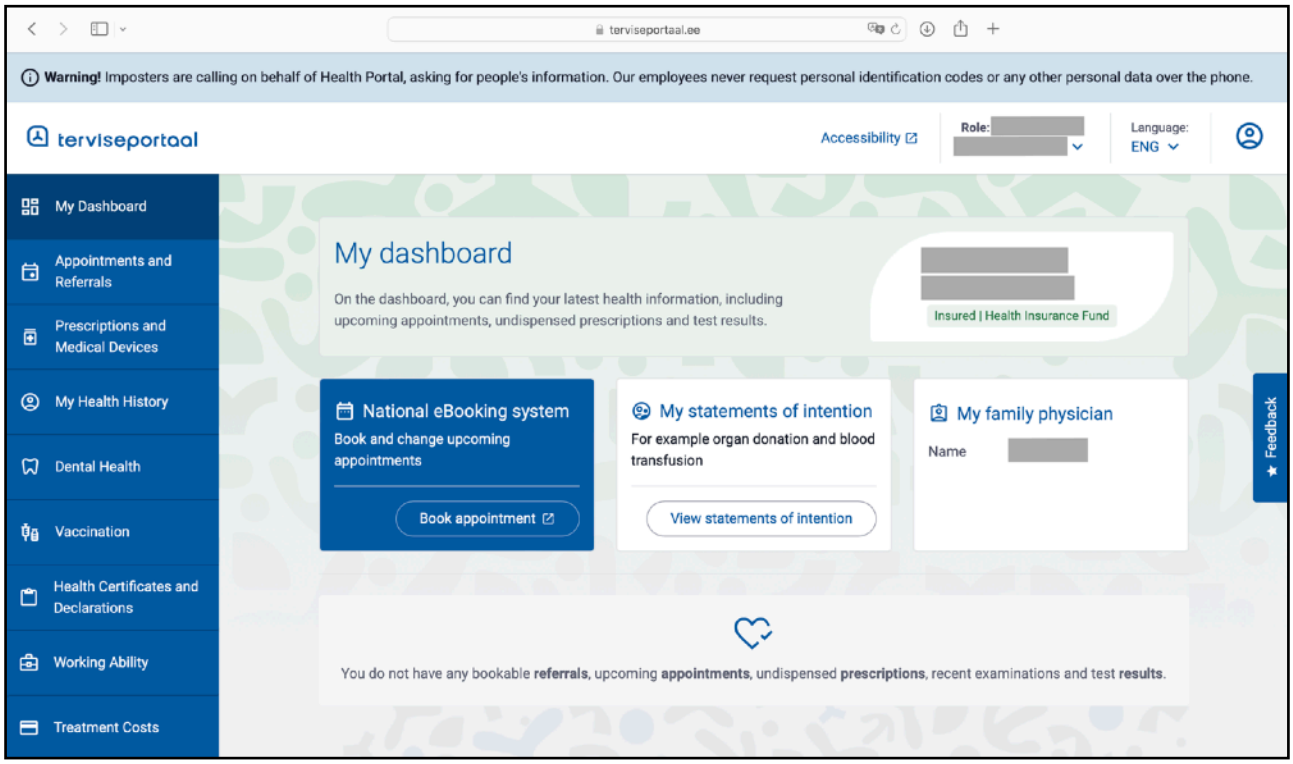


Figure 31: The dashboard view of the Health Portal. Sensitive information has been removed from the screenshot due to privacy concerns.

broader population, enhancing the overall landscape of healthcare options available to Estonians (Paraskevopoulos, 2022).

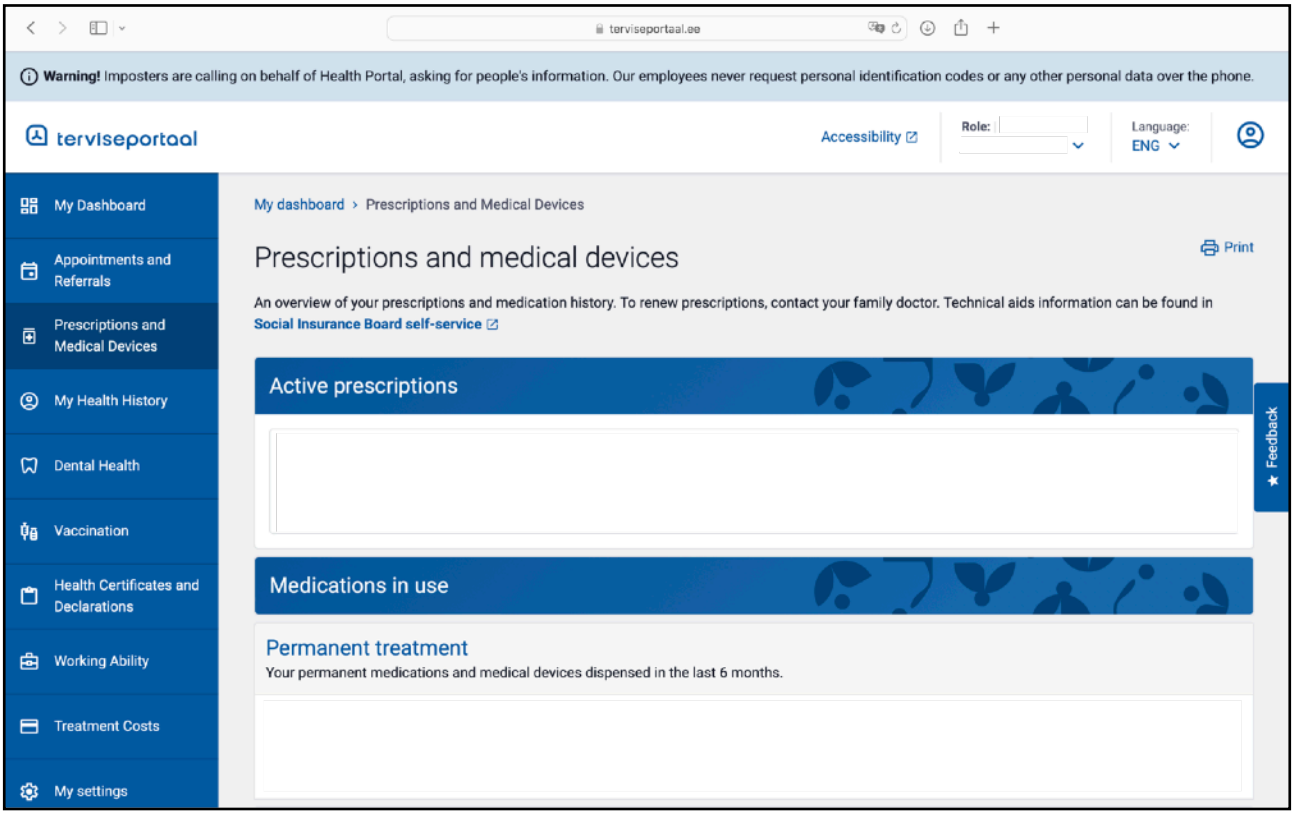


Figure 32: An example screen on the Health Portal, here showing the general structure of the Prescriptions and Medical Devices page. Sensitive information has been removed from the screenshot due to privacy concerns.

Analysis of the Health Portal

EHIF is the leading institution in managing the Health Portal platform. Three screenshots of its interface can be seen in Figures 30, 31, and 32. Terviseportaal, Estonia's national health portal, provides a centralized platform for citizens to access their health information, book appointments, and communicate with healthcare providers. The portal's design and functionality reflect a focus on e-health and digital innovation. The platform aligns with the nation's broader digital strategy, which emphasizes accessibility, user-centric design, and data security (Tiits et al., 2008). By utilizing its advanced IT infrastructure, Terviseportaal aims to enhance the efficiency and effectiveness of healthcare delivery in Estonia.

The website uses a combination of white, shades of gray, and blue. The blue color is often used in the healthcare domain since they are shown to be associated with trust, reliability, and professionalism (Su et al., 2019). This consistent color scheme is seen throughout the landing page (Figure 30), the dashboard (Figure 31), and the prescriptions and medical devices screen (Figure 32).The website uses a modern layout and typeface, thus focusing on readability and practicality. The left navigation bar highlights a wide variety of services that a user can access, ranging from health certificates and declarations to vaccinations to dental health.

Nationally, private healthcare providers in Estonia may offer their own digital solutions, but Terviseportaal has the advantage of being the official government-endorsed platform, ensuring wider acceptance and integration across healthcare services. (EHIF, 2024)
The primary users of the Health Portal are Estonian citizens and residents who seek convenient access to their health information and services. The portal is designed to cater to a diverse user base, including different age groups, tech-savvy individuals, and those with limited digital literacy. According to academic studies, user satisfaction in digital health portals is heavily influenced by ease of use, perceived usefulness, and trust (Venkatesh et al., 2012). The Health Portal's user-centric design, which includes clear navigation, accessibility features, and secure data handling, helps in meeting these expectations and building trust among users. Findings from the user study (Chapter 3.3.1 Context factors) indicate that the Health Portal is used infrequently, mainly for checking test results and setting doctor's appointments for both her and her children. The users indicated difficulty in understanding medical jargon and occasionally not receiving notifications about test results. Overall, however, e-state solutions, including the Health Portal, are trusted by interview respondents.

Conclusions from the two 4C analyses

The brands of the Estonian Health Insurance Fund (EHIF) and its Health Portal are focused on health-related services. EHIF is responsible for organizing national screening programs (among others), while the Health Portal plays a crucial role in communicating medical results to individuals. Consequently, the perception of the services provided by EHIF and the Health Portal is closely linked to their reputations and the trust they have built within the community.
However, the PRS presents a unique kind of medical result that stands apart from traditional health assessments. Unlike conventional medical results, which typically indicate the presence or absence of a specific condition or disease, PRS is designed to predict the potential for future health developments based on genetic information. This focus on the prediction of PRS results introduces a new situation that needs to be understood by users.
There is a significant risk that the PRS results may be misinterpreted as immediate indicators of a person's current health status. Given that the perception of EHIF and the Health Portal is connected with providing concrete health-related medical information, there is a possibility that individuals might place PRS results in the same category as other medical outcomes that have a direct and immediate impact on their health. If a PRS result is perceived similarly to other health results, including mammography screening results, annual health checkups, and results from occupational health checkups, the recipient likely perceives them as directly affecting her health. This could lead to confusion and unnecessary anxiety, as people might

not fully understand the probabilistic and predictive nature of PRS, mistaking it for a definitive diagnosis.

It is important to communicate the purpose and implications of PRS results clearly and effectively to mitigate this risk. By emphasizing that PRS is a tool for understanding potential future health risks rather than a diagnostic tool, EHIF, and the Health Portal can help individuals better interpret their results and take appropriate steps toward preventive health measures without excess concern.

The subsequent design process focused on framing genetic risk as a unique type of health insight with the goal of distancing it from health results with an immediate impact. On a perception level about the service, this first and foremost relates to the design concept being easy to understand for a variety of numeracy levels, PRS framed as a small change and an early warning, and that the person does not overstate the health implications of the service (relating to design criteria 1–4, respectively).

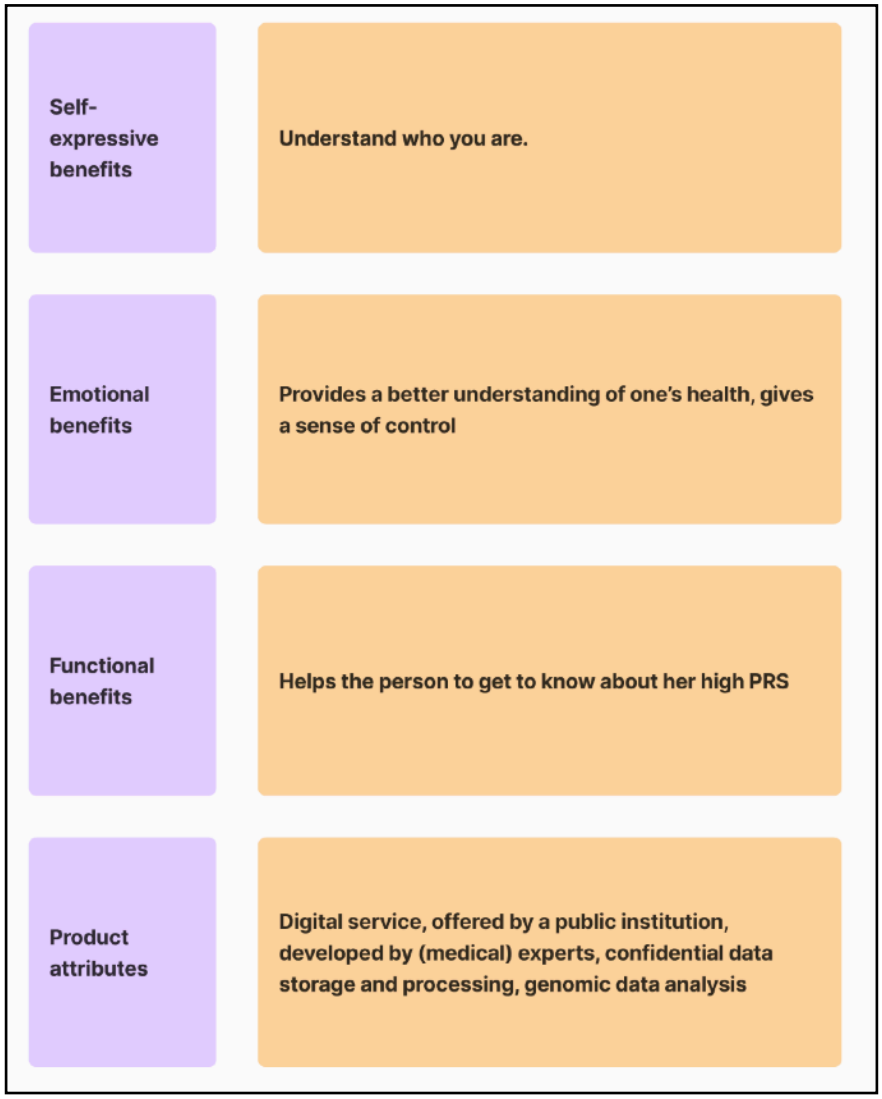


Figure 33: A brand ladder method (Wansink, 2003; In combination with the format from the "Brand and Product Commercialization" course at IDE) used in the process to arrive at the branding concept.

As a result of the brand ladder (see Figure 33; Wansink, 2003; in combination with the format from the "Brand and Product Commercialization" course at IDE) and a category migration method (from the "Brand and Product Commercialization" course), the concept of My Genetic Mirror is proposed. For this, guidelines for genomic counseling were analyzed from the literature review and the user interviews. The subsequent positioning focused on creating a different perception of the program. The concept aims to frame genetic health as a reflection of the person's identity, hence the "Mirror". In highlighting the concept of genomic risk, the concept focuses on communicating that there are both positive and negative aspects to everyone's genome.
As everyone's genome has positive and negative factors, the concept highlights what is unique for the person in their genome. The concept highlights connection ("My genome is part of who I am") and ownership ("This genomic data belongs to me"). If a person considers the situation theirs, they are more likely to take action, e.g., in taking part in screening in case of elevated risk. The overall concept thus emphasizes that "This is my situation, this is my identity". An alternative name variant "My Genetic Identity" for the service was considered, which was however discarded due to running the risk of the service being linked too closely with genetic ancestry services like 23andMe.

The connection between the fire safety and mirror metaphors

The mirror in My Genetic Mirror can be understood as a metaphor since it "reflects back" about the person's genetic health. Next to the mirror metaphor, the fire safety metaphor provides a mental model about genetic risk and the role of the PRS communications service. Together, the mirror and fire safety metaphors combine creating an understanding about genetic risk by using a tangible item that the recipient can easily imagine (a mirror) as a means to giving an overview about the person's genetic health.

"Beta version" as a first indicator of the service's limitations

The branding can be used to also communicate the limitations of the PRS results. As Interviewee 14 (expert) noted, people may perceive the PRS result as the full risk, which it is not. Since PRS is only a part of the risk (and for more comprehensive risk communication, further methods should be included, e.g. monogenic risk). To ensure that the recipient inherently perceives the result as not fully comprehensive, the overall service should be presented and handled as a beta version. In software, beta versions relate to unfinished programs or applications. Thus, as long as PRS is exclusively used for determining the risk level, a "beta version" label can give the first hint that the service is not yet finished, i.e. does not take the full risk into account. Simultaneously, however, it communicates that the results can be used for drawing first conclusions, but are not definite. Once further methods for risk

determination, e.g., monogenic risk, are included, the "beta version" label can be removed. As the cost of genetic testing is expected to decrease in the coming years, additional methods for risk determination can eventually be used.

Brand DNA

Brand DNA is a framework that characterizes a set of parameters that distinguish a company from its competitors, encompassing both external and internal characteristics, advantages, and the brand's essence conveyed to consumers (Maltuz et al., 2023). To formulate the brand DNA, the purpose, positioning statement, and personality were to be determined. For this, the content from the Master's course "Brand and Product Commercialization" was used. The first step was to determine whether a brand archetype could fit the personality part of My Genetic Mirror. Three candidate archetypes were chosen: the Sage, the Innocent, and the Caregiver (Table 10).

Brand archetype	Emphasis	Goal	Mantra
The Sage	data analysis, learning, research, and planning	Understand processes and underlying reasons	"The truth will set you free"
The Innocent	happiness, optimism, and simplicity	Desire for purity and positivity, often seeking to avoid negative outcomes	"Free to be you and me"
The Caregiver	support, safety, nurturing, compassion, generosity	Protect people from harm	"Love your neighbor as yourself"

Table 10: This table highlights the different brand archetypes under consideration for the Brand DNA.

Applied to the proposed design concept, the Sage personality indicates an approach focused on informational support and a limited focus on social support. Results from user research indicated that the recipients wish to avoid jargon and complex information, and the Sage archetype may put an excessively heavy informational focus on the communication. Thus, the Sage archetype was excluded.

The Innocent archetype follows the mantra "Free to be you and me", which is connected to the "identification" aspect of the design concept. The Innocent archetype is driven by a desire for purity and positivity, often seeking to avoid negative outcomes. In the context of the service concept, this archetype can help frame a negative PRS result in a way that emphasizes learning and growth, rather than as a setback.

The Caregiver archetype is often used for healthcare purposes. However, since the aim of the service repositioning is distancing from other health results, the Caregiver archetype may actually position it more among health results.
Although both the Innocent and the Caregiver archetypes were considered as good candidates, the Innocent archetype was ultimately chosen due to the possibility of more clearly distancing the service from other health results. Nonetheless, some elements of the Caregiver archetype were included, such as the focus on support and safety. As a result, the brand personality was constructed as an Innocent-Caregiver hybrid.
The positioning statement was formulated as the following:

> For 40-year-old Estonian women, My Genetic Mirror offers a digital health management service with high medical expertise that gives people the feeling of ownership and connection to identify with their own self.

This positioning statement focuses on the first horizon in the roadmap. For horizons 2 and 3 of the roadmap (see chapter 4.7), the target group "40-year-old Estonian women" should be expanded to include the wider population for whom the service is intended.
The third part of a Brand DNA focuses on the formulation of the purpose. It aims to answer the questions "Why did we start this? What do we add to the world?" in a short manner. For My Genetic Mirror, the Purpose was formulated as:

> Like Yin and Yang, genetic risk is a combination of the good and the bad.

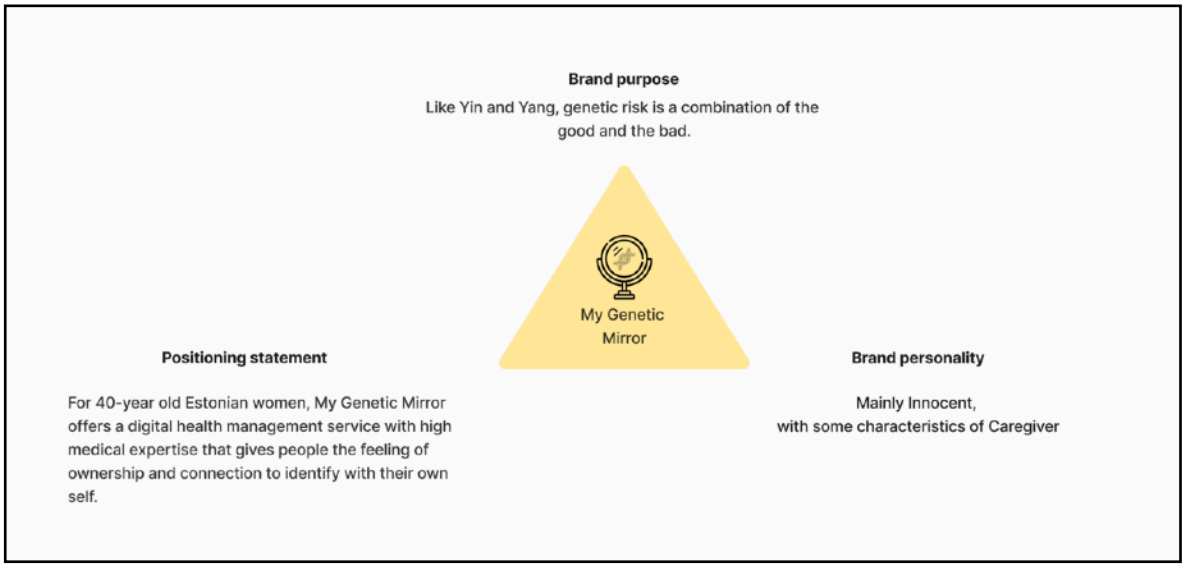


Figure 34: The Brand DNA for My Genetic Mirror.

The resulting visualization of the Brand DNA is formulated in Figure 34.

Simple brand guidelines for My Genetic Mirror

The brand guidelines of My Genetic Mirror reflect the characteristics of the Innocent brand archetype. Research by Elliot and Maier (2014) on color psychology explains how colors can influence perceptions and emotions. Soft, pastel colors are associated with calmness and positivity, which align with the Innocent archetype's values (Elliot & Maier, 2014). Since the purpose of the brand is to convey genetic risk as something with both positive and negative sides, light shades of yellow and red are used. Light yellow should be used where a positive

Figure 35: A draft poster for advertising the My Genetic Mirror service.

insight is communicated, whereas light red should be used for instances where the outcome can be perceived as negative or the user should pay attention to a given section. Since the Innocent brand archetype relates to simplicity, a minimalistic sans-serif font is used. The resulting brand guidelines were employed in a draft poster for advertising the service (Figure 35).

4.7 Roadmapping

This chapter shows the results of the Creative Trend Research process (Simonse, 2018), highlights the future vision for 2030, and goes through the three horizons for both the strategic and the tactical roadmap. The first horizon focuses on communicating BC PRS results. The second horizon focuses on preparing for integrating multiple PRS results into holistic risk communication. The third horizon focuses on providing a personalized understanding of the person's genetic health via utilizing an AI virtual genetic counselor via video call. The strategic and tactical roadmaps are split into three horizons, each two years long, between 2024 and 2029 (for the time pacing strategy, refer to Chapter 4.7.3).

4.7.1 Creative trend research

Based on the trend research using the Trend Patterns method (Simonse, 2018; Figure 36), I identified and clustered several key trends that are crucial for shaping future healthcare strategies. The clusters, marked in yellow, and their specific aspects, indicated in purple, are described as follows.

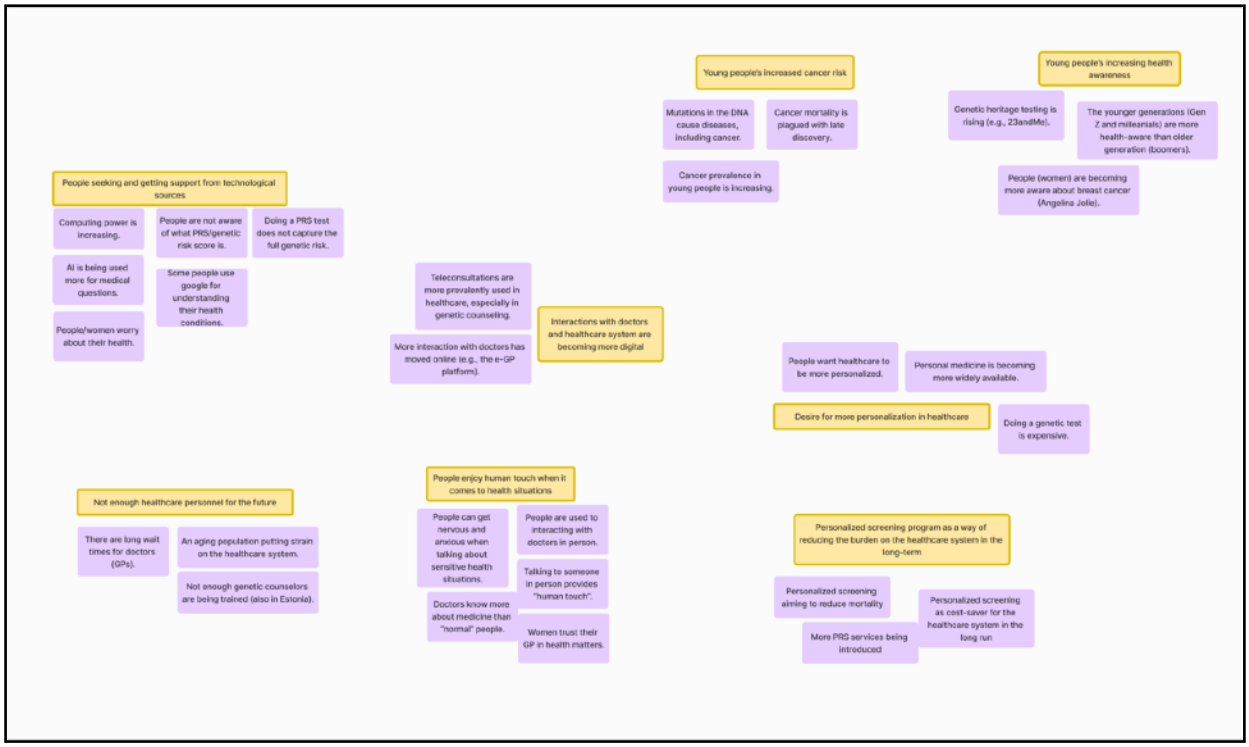


Figure 36: A synthesis of the collected insights from the context matter and according to the Trend Patterns technique.

1) People seeking and getting support from technological sources

This cluster focuses on the increasing reliance on technology for health-related support and information. Key aspects include rising computing power, the use of AI for medical questions, and people worrying about their health. There is also a noted lack of awareness about PRS (genetic risk scores), with some individuals using Google to understand their health conditions.

2) Young people's increased cancer risk

This trend highlights the growing concern over cancer among young people. Aspects include the increase in DNA mutations causing diseases, the late discovery of cancer leading to higher mortality, and the overall increase in cancer prevalence among the young population.

3) Young people's increasing health awareness

Younger generations, particularly Gen Z and millennials, are becoming more health-aware. Genetic heritage testing is rising, and there is an increased awareness of specific health issues, such as breast cancer risk, influenced by high-profile celebrities like Angelina Jolie.

4) Not enough healthcare personnel for the future

This cluster addresses the shortage of healthcare professionals in Estonia. Aspects include long wait times for doctors (GPs), an aging population putting strain on the healthcare system, and a low number of genetic counselors in the country.

5) People enjoy human touch when it comes to health situations

Despite technological advancements, there is a strong preference for human interaction in healthcare. Aspects include people’s nervousness and anxiety when discussing sensitive health situations, the trust people place in their GPs, and the comfort derived from in-person consultations, which provide a "human touch."

6) Desire for more personalization in healthcare

There is a growing demand for personalized healthcare solutions. Personalized medicine is becoming more widely available, though genetic tests remain expensive. Personalized screening programs are seen as a way to reduce the burden on the healthcare system in the long term, potentially lowering mortality rates and serving as cost-savers. (Sun et al., 2022)

These findings suggest a significant shift towards digital and personalized healthcare solutions, driven by technological advancements and a growing awareness of health risks among younger populations. However, the trends also highlight challenges, such as the shortage of healthcare professionals and the need to maintain the human touch in health interactions. Overall, these insights highlight the need to integrate technology with personalized care to meet future healthcare demands.

4.7.2 Future visioning

According to the future visioning approach (Simonse, 2018), a future vision for 2030 was imagined and formulated. Using the insights gathered in the previous section, a desired future state was articulated. To arrive at the final formulation, multiple iterations were performed. The future vision was formulated as the following:

> Creating emotionally reassuring yet cost-efficient genetic health management for a peace of mind

In the future vision, each of the three values plays an important role for the future state. Emotional reassurance focuses on calming down a PRS recipient about the meaning and implications of the result. This addresses the emotional needs of users, making them feel safe and confident in understanding PRS. Cost-efficiency focuses on delivering value without incurring excessive strain on the healthcare system by leveraging digital communication tools, scalable educational resources, and automated scheduling for follow-ups. This ensures that users receive the necessary support and information in a financially sustainable manner. Genetic health management refers to a comprehensive approach to not just helping individuals to make sense of their genetic information, including genetic risks, at only one point in time (the digital PRS result communication), but also to enabling them to monitor and manage the risks over a prolonged period of time. Peace of mind relates to PRS recipients feeling reassured not just about their PRS results, but know that their overall genetic health is in good hands and under control. The path towards the future vision is highlighted in three distinct horizons, which are introduced and elaborated in the following.
First, the horizons of the strategic roadmap are discussed, which looks at the concept from a strategic lens, focusing on values, trends, and the general service proposition. Then the tactical roadmap is highlighted with relevant technologies, resources, and relevant insights into the components of the value proposition.

4.7.3 Time pacing strategy and technology scouting

Time pacing strategy

The strategic and tactical roadmaps are split into three horizons, each two years long, between 2024 and 2029. Once the service propositions were formulated during the conceptualization phase, the time pacing strategy was determined. For this, the determination started by estimating from which year, considering current technological progress and societal trends, the AI assistant genetic counselor would become feasible, viable, and desirable. Initial service propositions utilizing avatar-supported AI assistants, including for health advice, already exist and serve users (Soul Machines, 2022; Replika, n.d.; kuki_ai, n.d.). Thus, a noteworthy level of

feasibility and viability is present. These are expected to develop further regarding conversational abilities, emotional intelligence, and contextual understanding, enabling them to handle more complex interactions and tasks (Aslam, 2023; Taecharungroj, 2023). Although AI chatbots are being already used now, with rising acceptance (Tian et al., 2023), I expect that AI assistants supported by an audiovisual avatar to start becoming more accepted in society during the next five years (relating to desirability). Thus, the third horizon was formulated for the year 2029. By backtracking the horizons to the present year (2024), three two-year-long horizons were determined. There, the first horizon focuses on incremental innovation, with the second horizon aiming to bridge the first horizon with the disruptive third horizon (Simonse, 2018). The horizons are elaborated in detail in the following sub-chapters.

Technology scouting

For technology scouting, the strategic partitioning approach (Simonse, 2018) was used to map the needs of different personas with technologies that can help the service proposition tend to those needs. The personas can be assigned into two clusters: the ones open to receiving a PRS result online (the Technologically Savvy, the Pragmatic, and the Indifferent) and the one looking for human touch in a follow-up consultation. Insights from mind-mapping sessions throughout the conceptualization process were used, along with gathering technology-related insights in parallel to Creative Trend Research. This resulted in proposing an AI chatbot for around-the-clock support to help clarify questions for the first cluster of personas. In tending to the second cluster, it was important to provide support in a way that can emulate human touch accessibly via a digital channel. There, AI assistants with audiovisual avatars were ultimately chosen. The AI chatbot and AI assistant are discussed in more detail in the following sub-chapters.

4.7.3 Strategic roadmap

Figure 37: Overview of the three horizons of the strategic roadmap.

An overview of the three horizons of the strategic roadmap is included in Figure 37. The following sub-chapters elaborate on the three horizons. The entire strategic roadmap is included in Figure 42.

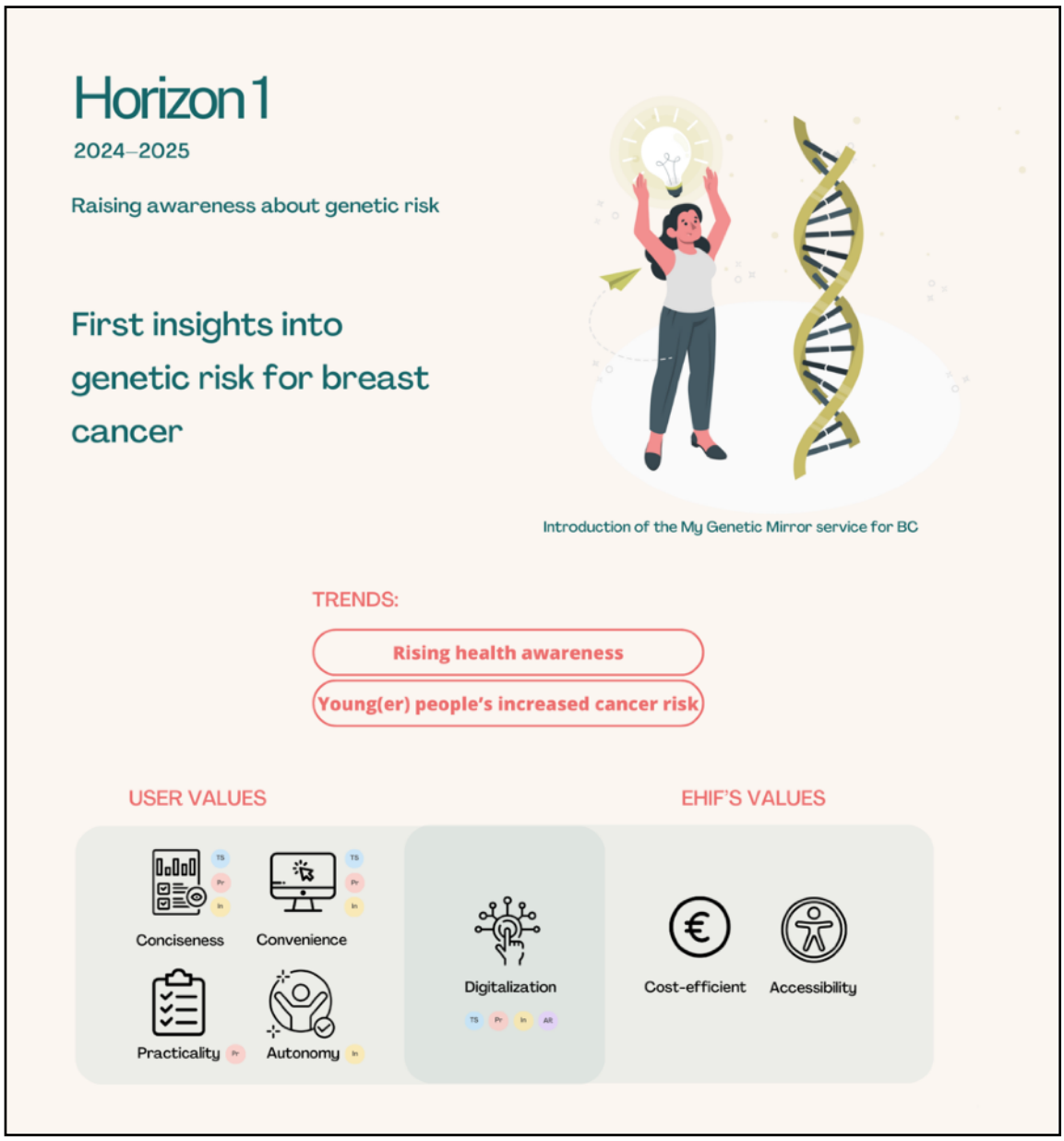


Figure 38: Overview of the first horizon of the strategic roadmap.

Horizon 1 (2024–2025)

The first horizon (Figure 38) focuses on raising awareness about genetic risk by first providing insights about future breast cancer risk. In this horizon, the main focus is on personas the Technically Savvy, the Pragmatic, and the Indifferent. This is because, as opposed to the Anxious Receiver, they are more self-management-focused, meaning that they can manage genetic risk information more independently, and are less likely to need emotional reassurance in an in-person consultation.

In Horizon 1, the values of the TS, Pr, and In include a need for concise information (all three personas), convenience of receiving and clarifying PRS information online (all three personas), the focus on practical next steps (the Pragmatic), and the need for autonomy (the Indifferent). On the EHIF side, they value cost-efficiency, accessibility, and digitalization. With a look at both the user side and the EHIF side, digitalization is valued. Digitalization here

refers to especially delivering PRS information via a digital channel, which the Anxious Receiver persona is open to.

Trends for this horizon highlight that younger women show an increase in breast cancer rates (Xu et al., 2024) and that health awareness is rising among the population (Tamson et al., 2022; Mikk et al., 2021). In this situation, My Genetic Mirror provides the first insights into BC PRS and the follow-up steps, if necessary. Here, a website format is utilized with an email invitation system that includes all of the components of the previously introduced service concept (see Chapter 4.6).

Horizon 2 (2026–2027)

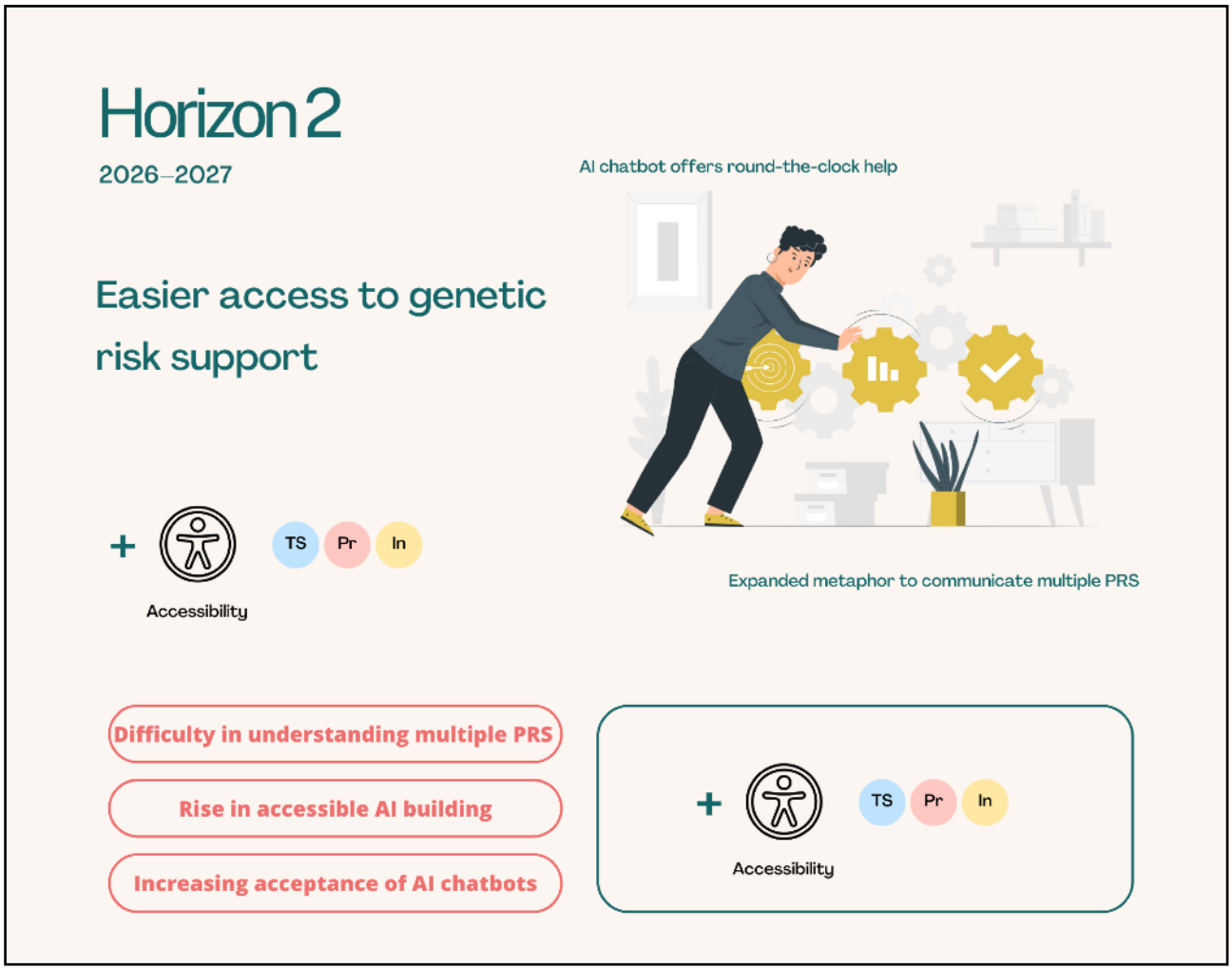


Figure 39: Overview of the second horizon of the strategic roadmap.

The second horizon (Figure 39) focuses on making genetic risk support more accessible. This is done in two ways: by expanding the mental model of the recipient and by making follow-up advice available round-the-clock. This comes in a time period where EHIF plans to introduce the digital communication of multiple PRS scores. Thus, the value of conciseness and accessibility for follow-up questions becomes ever-important for the TS, Pr, and In, since it is

important for them to understand their health situation in a simple way for multiple PRS. This horizon also overlaps with the trends of increasing access to building AI models and an increasing societal acceptance of AI chatbots, especially in healthcare.
In this horizon, the metaphor from horizon 1 is expanded to communicate a mental model for multiple PRS, and an AI chatbot is implemented to give instant feedback to follow-up questions (see the tactical roadmap for both).

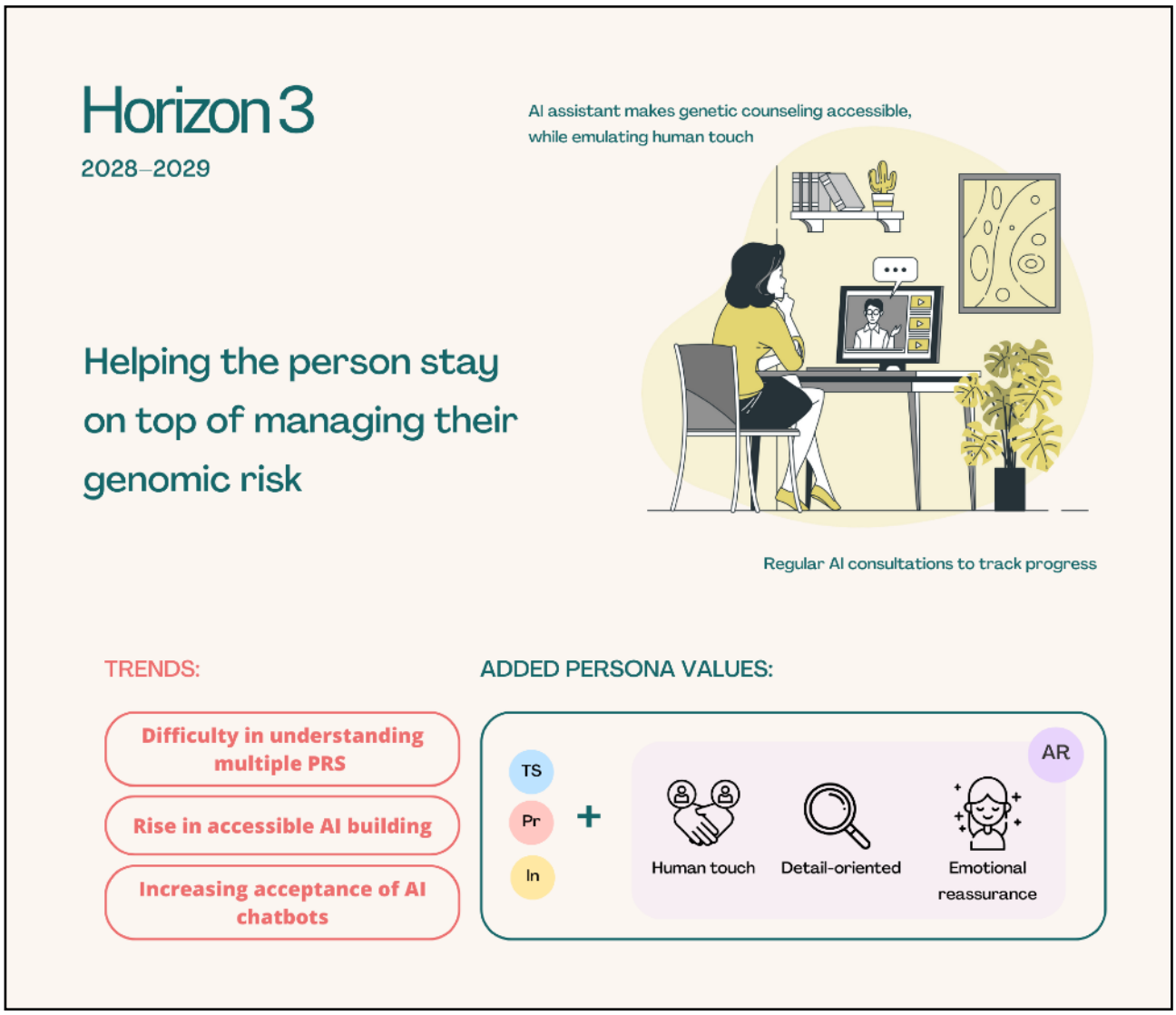


Figure 41: Overview of the second horizon of the strategic roadmap.

Horizon 3 (2028–2029)

The third horizon (Figure 41) focuses on helping the person stay on top of managing their genomic risk. In this time period, the shortage of healthcare personnel in Estonia is increasing, with longer waiting times. Simultaneously, however, PRS recipients, especially including the persona Anxious Receiver, still wish for a human touch in interactions with healthcare professionals. Moreover, the target group's acceptance of AI as a supporter of health self-management has increased.

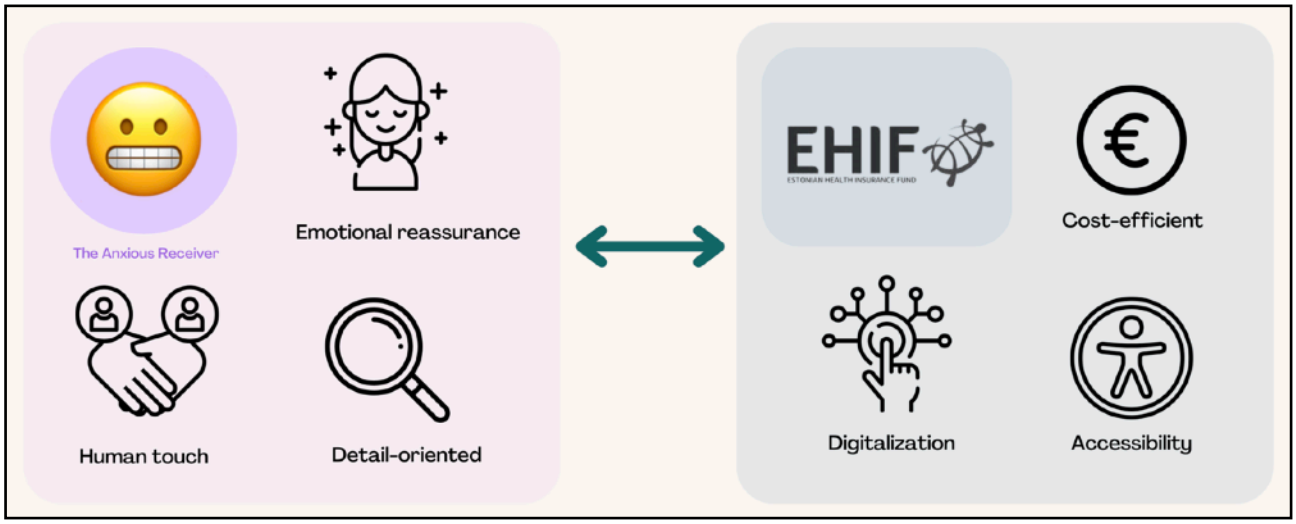


Figure 40: Value tension between the Anxious Receiver and EHIF.

Before the third horizon, there is a value tension between the Anxious Receiver and EHIF (Figure 40). On the one hand, the Anxious Receiver values emotional reassurance, human touch in PRS communication, and she is detail-oriented. She values access to healthcare professionals to understand the implications of her PRS result. On the other hand, EHIF focuses on cost-efficiency and digital accessibility to their services. For EHIF, consultations with healthcare professionals are relatively costly and they put strain on the healthcare system. Due to this reason, the Anxious Receiver's values have only been included partially for initial digital communication of the score, but offering the persona the possibility to consult a healthcare professional.

In horizon 3, the values of the Anxious Receiver are incorporated. To support the AR in getting assistance in a time of long queues to medical professionals, the human touch is being emulated via an AI assistant, supported by an audiovisual avatar. It is expected that by 2028, the technological development of AI avatars will advance at a fast pace, bringing more advanced human-like features to give an impression to the recipient as if they are talking to another human. This approach aims to bring genetic counseling to the recipient's digital devices, enabling access to on-demand assistance. The PRS recipient can interact with the AI assistant both before taking part in the service and after the result communication, helping the person to be supported along the user journey.

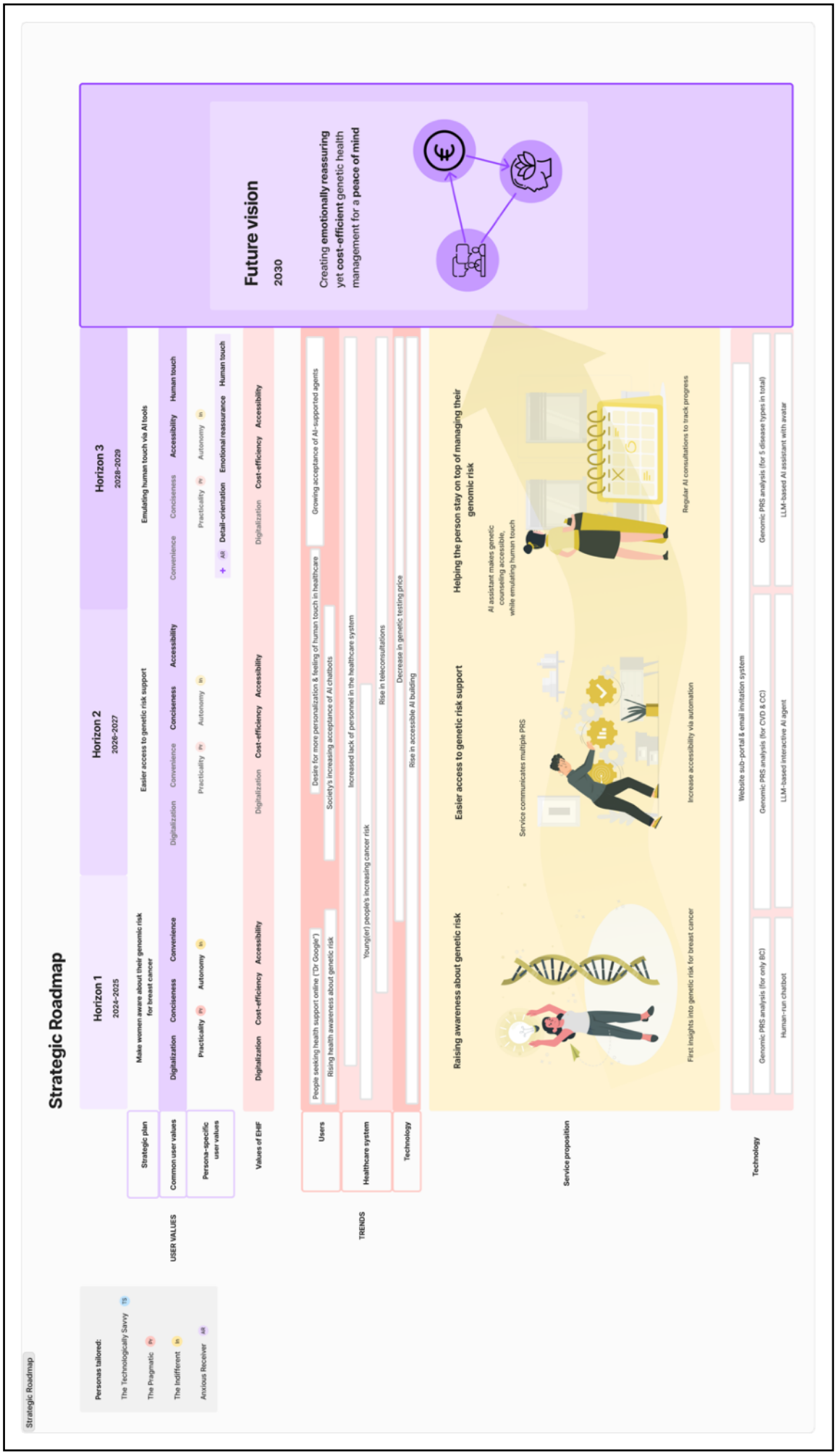


Figure 42: The resulting strategic roadmap.

Furthermore, since the AR also aims for regular consultations to know her progress in managing the genetic risk, the PRS recipient is prompted with the option to schedule regular online AI consultations to determine her progress.

4.7.4 Tactical roadmap

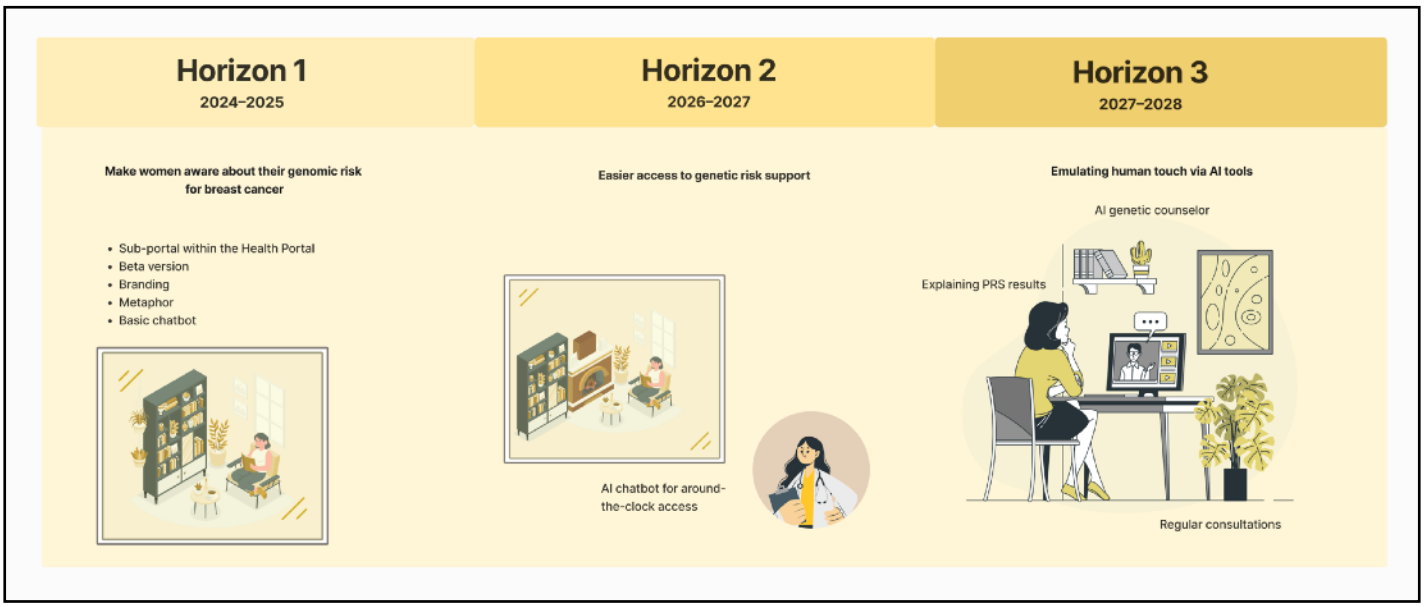


Figure 43: Overview of the three horizons of the tactical roadmap.

Figure 43 provides an overview of the three horizons of the tactical roadmap. Figure 44 highlights the different technologies relating to the three horizons, which are further elaborated in the following three sub-chapters. Figure 48 shows the resulting tactical roadmap.

Horizon 1 (2024–2025)

The first horizon focuses on communicating a single type of PRS result – for BC. Here, two central concepts are introduced: My Genetic Mirror and the fire safety metaphor. The former focuses on general framing of the service whereas the latter focuses on specifically communicating PRS risk via a metaphor. To support the perception of the service, the proposed Brand DNA should be implemented (Chapter 4.6).

Since communicating PRS requires a level of personalization, the first initiatives are being started in Horizon 1. First, a basic human-powered chatbot assists in case of follow-up questions. If a person wants a consultation, she is provided with the necessary contacts to a genetics-focused specialist.

Several resources are necessary for reaching the goals of this horizon. First, a collaboration with the Health and Welfare Information Systems Centre (TEHIK) should be initiated to create the My Genetic Mirror section on the Health Portal. Secondly, an AI development team should be established to develop and test the AI chatbot. Thirdly, a research group should be established to study PRS communication for the wider population, including men. This serves as preparation for the second horizon, where further PRS testing (in addition to BC) is planned to be introduced. The goal of the research group is to understand the kind of support that is necessary for eventually communicating PRS results to the wider population. The

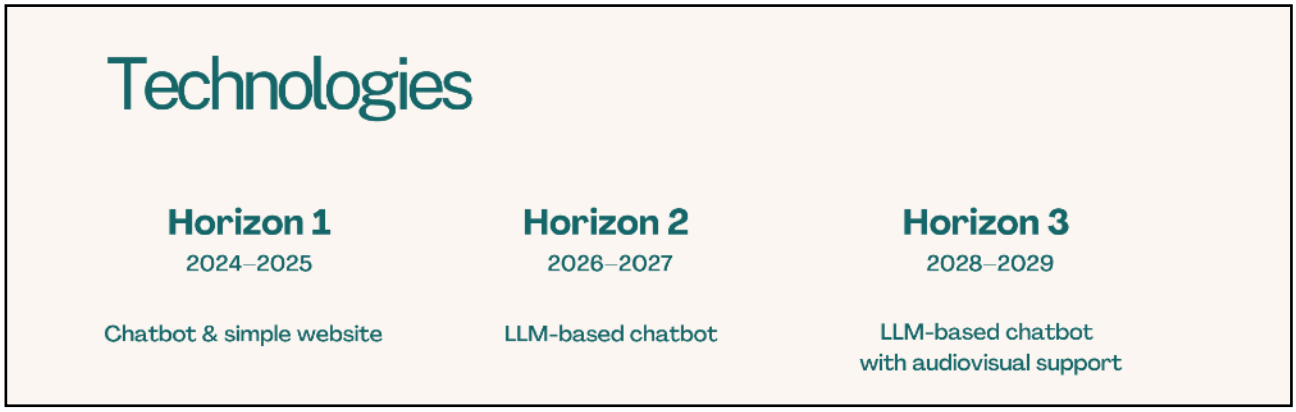


Figure 44: Overview of the technologies by horizon.

resulting insights can be used for adjusting and tailoring the PRS communication for Horizon 2. The research work should be finished by the end of Horizon 1. Qualitative evaluation can be performed from the questions on the chatbot.

Horizon 2 (2026–2027)

The second horizon focuses on advancing the concept of My Genetic Mirror for communicating multiple PRS. During this stage, the users will receive separate PRS results for multiple diseases. However, this inherently complicates the communication for the recipient since she needs to make conclusions about her health based on multiple results. Here, an expanded fire safety metaphor should be implemented.

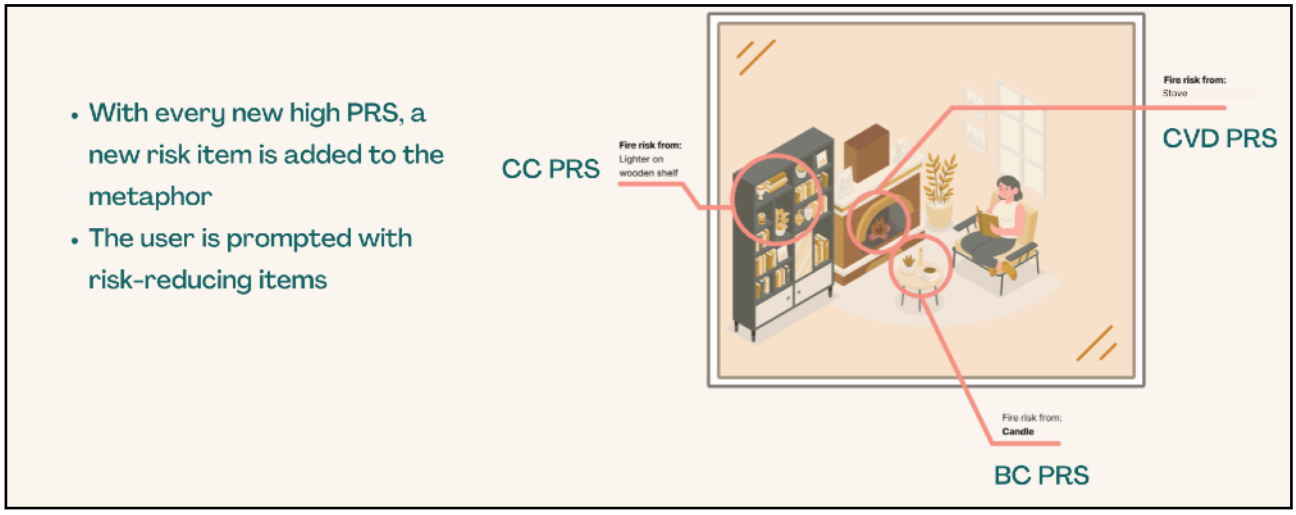


Figure 45. Overview of the expanded metaphor, including multiple PRS risks.

To support the perception of the service, the proposed Brand DNA (chapter 4.4.6) should be expanded to include further target groups of the service. The My Genetic Mirror page should include a module that helps to summarize multiple PRS results. These texts would be human-generated by genetic experts for different risk-level combinations. The expanded fire safety

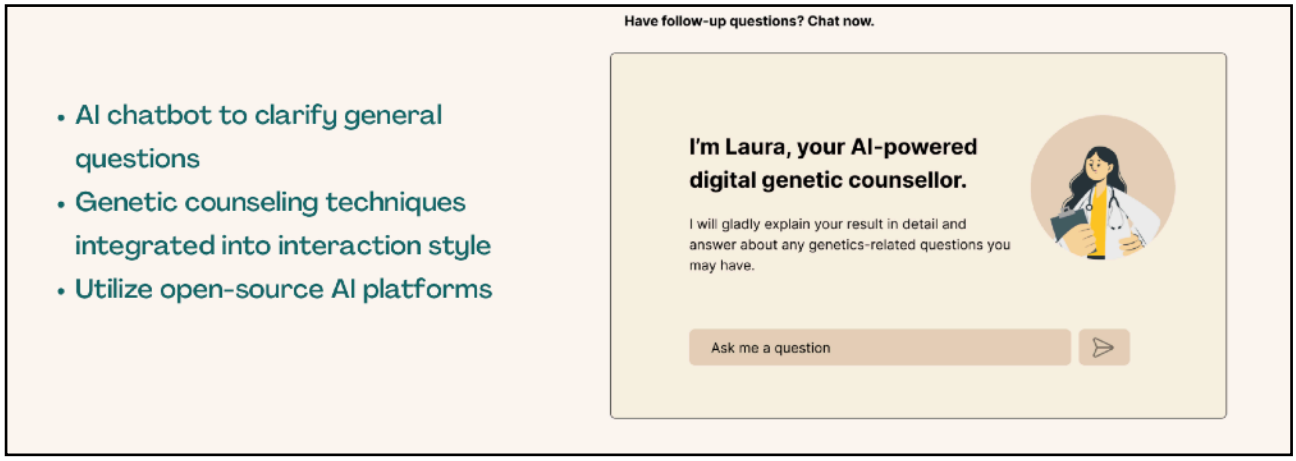


Figure 46: Overview of the AI chatbot utilized in horizon 2.

metaphor reflects the addition of further PRS results into the risk communication. The additional PRS results are incorporated into the expanded metaphor (Figure 45). With every new high PRS result, a risk item is added, e.g., a candle, a stove, a lighter on a wooden shelf. Thus, the recipient is prompted with risk-reducing items, such as a fire extinguisher, a smoke detector, a fire break to keep the risk low. If the person takes part in screening, the risk-reducing item is placed on the mirror view. The expanded metaphor thus aims to motivate users to take action about their health via screening program participation in order to minimize their risks.

During the second horizon, the development and testing of an AI-enabled chatbot (Figure 46) should reach the point when it can be implemented in the PRS results delivery. This will free up the genomic expert from answering follow-up questions in the existing chatbot and will enable questions to be answered immediately and with tested accuracy. After the introduction of the AI chatbot, monitoring and following fixes will be performed based on collected user data, for which consent is requested. The chatbot will be introduced as a personified virtual assistant with a genomic counseling specialization.

During Horizon 2, research conclusions into communicating PRS results to the wider population, including men, should be implemented into the service. This will be required for introducing further PRS communication, e.g., for cardiovascular disease (CVD), where men also comprise the target group.

Horizon 3 (2028–2029)

During the third horizon (2028-2029), the focus shifts to delivering highly personalized and streamlined genetic risk management through advanced digital communication technologies. By this time, video-based conversational AI models are anticipated to have gained widespread acceptance among the target group, facilitating the integration of AI-driven consultations into the genetic counseling process. This horizon aims to enhance the user experience by offering

more interactive and immediate support and reducing the burden on human genetic counselors.

The service will introduce an AI agent specialized in genetic counseling, capable of conducting short video call consultations, providing users with personalized advice and explanations based on their PRS results. The AI assistant (Figure 47) will serve two primary purposes: firstly, to explain PRS results in a manner that aims to emulate human touch, offering a more accessible service that delivers detailed and empathetic responses to user queries. Secondly, insights from expert interviews highlighted the importance of adhering to lifestyle changes and periodic monitoring, noting that PRS recipients often forget about their risk over time. Therefore, the AI assistant will function as a periodic check-up tool, enabling users to receive regular updates on their progress with lifestyle changes and monitoring efforts. This dual

Figure 47: Mock-up interface for the audiovisual AI assistant with a virtual avatar.

functionality ensures that users not only understand their genetic risk but also stay engaged in proactive health management.

The integration of AI-driven consultations will increase accessibility by providing around-the-clock access to genetic counseling, offering immediate support to reduce anxiety and uncertainty, alleviating the workload of human genetic counselors by handling routine follow-up consultations, and enhancing user engagement through interactive video call consultations.

To achieve believable training of AI assistants and ensuring peace of mind for users, the AI system should incorporate several key elements. First, transparency in AI training methods and data sources is crucial. Users should be informed about how the AI is trained, including

the ethical guidelines and data privacy measures in place. This transparency builds trust and reassures users about the safety and reliability of the AI. Second, incorporating learnings from user interactions with the chatbots from Horizons 1 and 2 should be utilized to train the AI model. Furthermore, advanced natural language processing and emotional recognition technologies that allow the AI to respond in a supportive and understanding manner should be utilized to react to users' needs. Additionally, the AI should facilitate easy access to human professionals when needed, ensuring that users always have the option for a personal touch in more complex or sensitive situations. Finally, continuous improvement based on user feedback and rigorous testing ensures that the AI remains effective and responsive to user needs.

Horizon 3 also anticipates the introduction of additional PRS tests, expanding the scope of genetic analysis to include a total of five different PRS assessments. This expansion will enable a more comprehensive understanding of an individual's genetic risk profile, covering a broader range of conditions. To help accommodate this increase in information, the service will also evolve the fire safety metaphor to effectively communicate the expanded insights from PRS results.

Despite the advancements in AI and digital communication, enabling participants the opportunity to reach out to an in-person consultation remains important, especially for the Anxious Receiver. This approach aims to balance the efficiency and accessibility of AI with the empathy and personalization of human interactions. Personalized notifications will keep users engaged and proactive in managing their genetic risks, and the AI system will be designed to recognize situations where human intervention is necessary, ensuring users receive the appropriate level of care and support.

The successful implementation of Horizon 3 will rely on several technological advancements and integrations, including the continued development of AI algorithms to improve the accuracy and responsiveness of the AI counselor, and robust data security measures to protect user privacy. This includes moving the consultation to an on-device setting, meaning that any sensitive information that the person communicates to the AI will remain on-device. The sensitive information mentioned in the consultation will be deleted afterward. By avoiding the transfer of data into servers that are beyond the user's control, the service will protect user data.

Expansion of the digital PRS result communication during Horizons 2 & 3

The My Genetic Mirror page on the Health Portal is set to be expanded during Horizon 2 and 3 to accommodate further PRS results and to provide a holistic interpretation that combines the results from the different PRS analyses. These results are combined with an expanded fire safety metaphor that aims to simplify the understanding of multiple PRS results at once. In

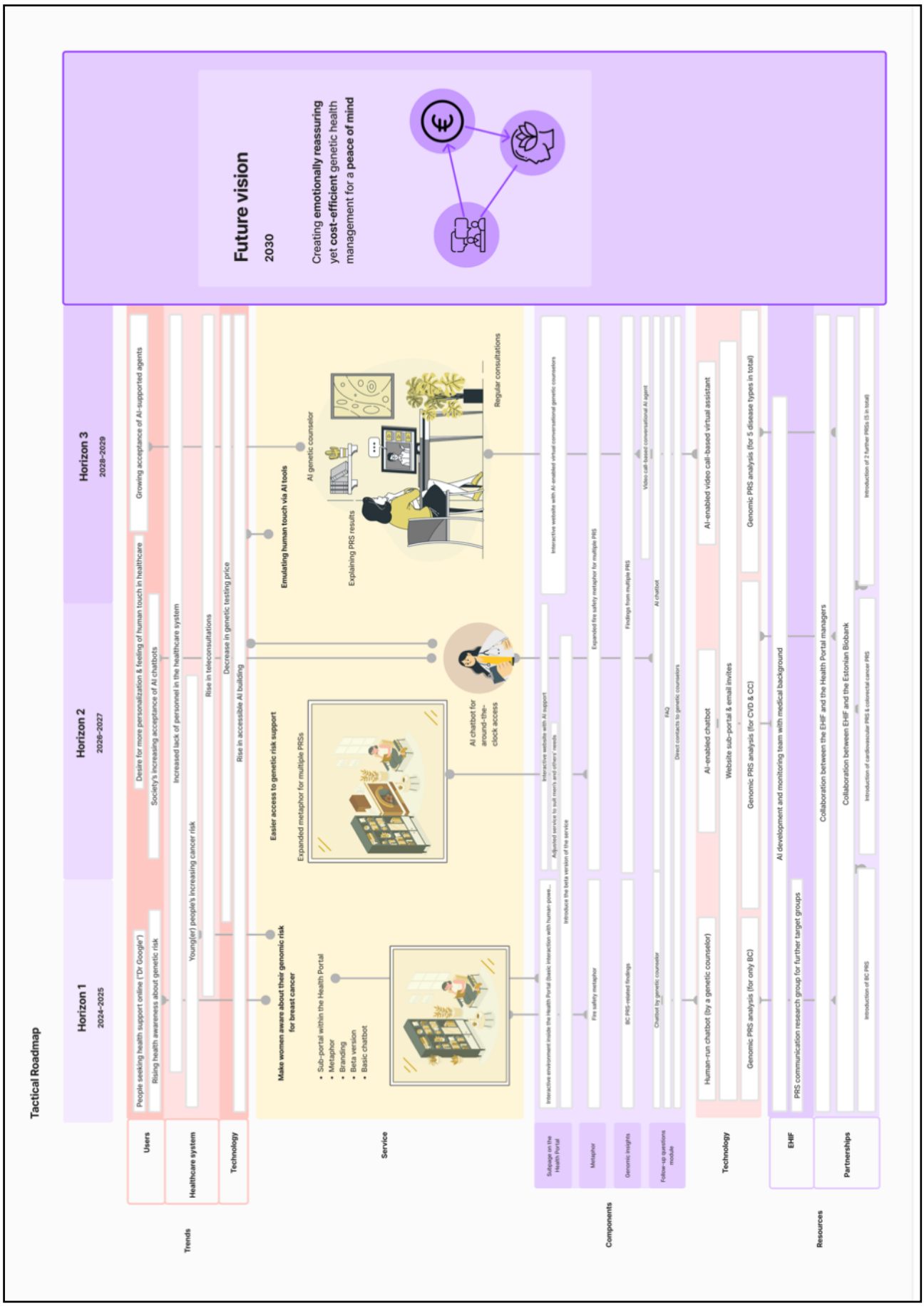


Figure 48: The resulting tactical roadmap.

horizon 1, the mirror view into the person's home shows a candle, which is highlighted as a risk for fire, and for which the smoke detector serves as a fire safety precaution. In further horizons, each new PRS type gets its own item that is understood as a fire risk. For example, an electric heater, a stove, or a lighter. For each newly added fire risk item (serving as analogies for different PRS types), a fire precaution item is provided (e.g., a fire extinguisher, a firebreak in front of a stove, a mini-grip plastic bag for the lighter), serving as metaphors to ways one can protect herself/himself from cancer and other types of disease. This approach aims to make receipt of multiple PRS results easier to grasp for the user.

5. Conclusion

This chapter wraps up the thesis. The following subchapters discuss the light evaluation, the relevance of the results, the limitations, recommendations, and related comments. Then, a personal reflection highlights the author's thoughts on the process, the results, and the project as a whole. Finally, a summary concludes the chapter.

5.1 Light evaluation

5.2 Contribution to new knowledge

5.3 Limitations, recommendations, and comments

5.4 Personal reflection

5.5 Summary

5.1 Light evaluation

5.1.1 Rationale and objective

Rationale

The light evaluation serves multiple purposes: to gather first feedback about the design concept, to serve as input for improving the concept as well for further suggestions. The evaluation focused on an interactive prototype of the service concept based on the service blueprint. The service concept relates to both the service blueprint as well as the first horizon of the strategic and tactical roadmaps, thus by including these in the light evaluation a substantial part of the design concept was considered. Another reason relates to practicalities, since the design concept is proposed to EHIF and the service blueprint is proposed for immediate implementation, an evaluation of the service blueprint will provide insights for EHIF into how potential users would react the design concept. For the light evaluation, a product usability evaluation based on Van Boeijen et al. (2014) was conducted.

Objective

The concept usability session serves as a first light evaluation of the concept. The objective of this usability evaluation is to assess how well the interactive prototype of the "My Genetic Mirror" service concept responds to user needs and influences their behavior, particularly focusing on explaining the concept of genetic risk, PRS, and the subsequent result, highlighting follow-up actions, and avoiding anxiety and worry throughout the user journey.

5.1.2 Presumptions

For the concept usability evaluation stage, four presumptions were formulated. They are highlighted in the following:

- The fire safety metaphor component should aid the user in understanding the concept of genetic risk and the role of the My Genetic Mirror service.
- The Follow-Up Questions Module should help the user clarify questions and concerns about the service and the result.
- The branding concept of My Genetic Mirror should distance the perception away from health results to framing genetic risk as part of the person's identity and thus calm down recipients.
- The spread-out information delivery should help to avoid an overwhelming delivery of information about genetic risk.

5.1.3 Research questions

The evaluation was based on the original research question "What informational and social support do women receiving a polygenic risk score for BC need in the context of a digital communication tool?" (Chapter 3.1) and the design criteria (Chapter 4.4).
The following research questions were formulated:

- What are the users' emotional responses to the provided information (throughout the user journey)?
- Are users likely to take action as a result of the information provided? What kind of action?
- How easily can users navigate through the online demo?
- What do the users consider the most and least calming elements of the service concept?

5.1.4 Participants and procedure

For the light concept evaluation, four IDE students were recruited. The sampling process followed a convenience sample of women between the ages 30–40 with no previous breast cancer experience, aiming to somewhat overlap with the age group of the participants from the user study. The resulting sample of design students consisted of women between 30–33. It was noted that the younger sample does not properly overlap with the target group from the user study, however, for a first understanding of the concept from the users' perspective, I considered the sample still useful for drawing first conclusions.
The light evaluation sessions were conducted individually and ranged between 30-45 minutes. The evaluation was done on a laptop and formulated as a prototype on Figma, enabling users to go through the general user pathway of the service, starting from the invitation email to the service and ending with the result communication page. The prototype is introduced in the video showcase provided with the thesis.
The participants were encouraged to "think out loud" as they were going through the prototype. Afterward, the reformulated research questions were posed to the participants. Finally, the users were asked to draw out an Emotional Journey (Open Practice Library, 2022) as a curve along the main service stages on a simple calm-to-anxious scale with the goal of measuring the participant's emotional state. User consent was obtained for audio and screen recording along with written notes. The collected data was pseudonymized, e.g., with "E1" referring to the first participant in the evaluation, and ending with "E4" (the fourth participant).

5.1.5 Results of the light evaluation

Participants' Emotional Journey curves

This sub-chapter highlights the different Emotional Journeys drawn by the participants (and digitally recreated by the author of this thesis) at the end of the light evaluation sessions. The participants experienced the service in different ways, ranging from more fluctuating (E3) to more stable emotional states (E2). Furthermore, the start and end node positions varied. The following highlights the emotional journey of each participant in further detail.

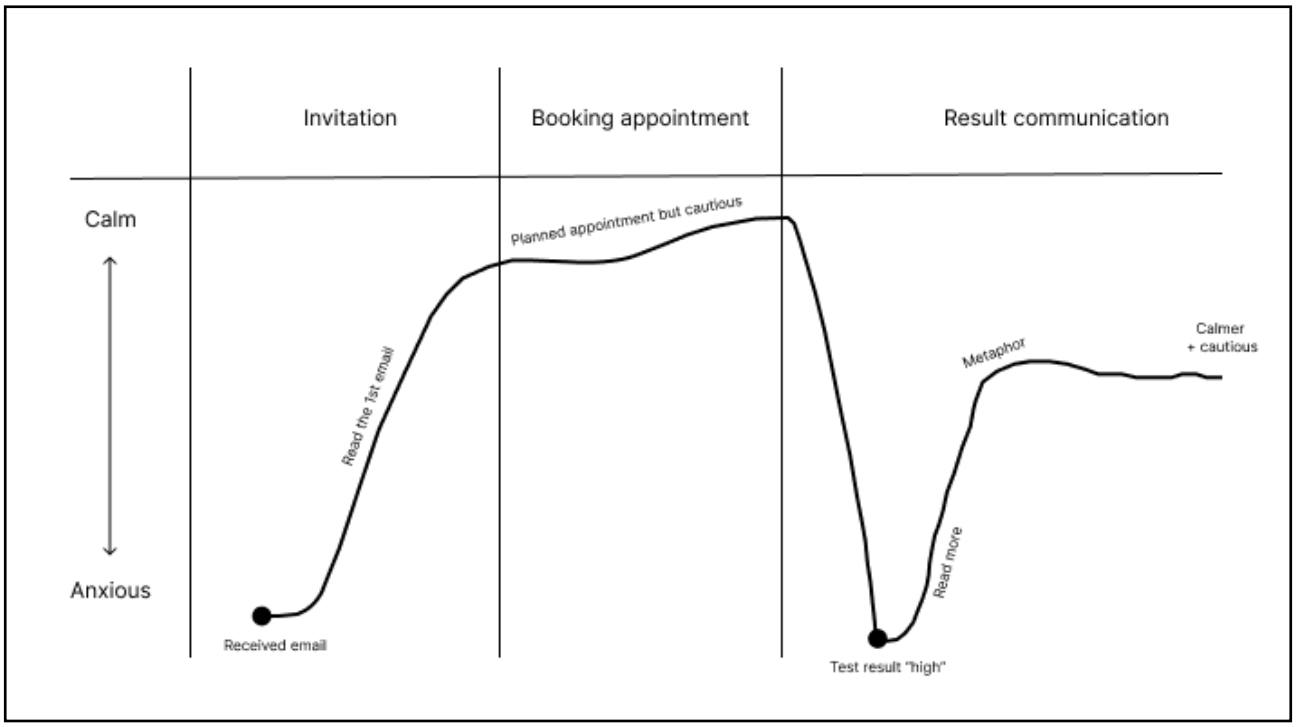


Figure 49: The Emotional Journey (Open Practice Library, 2022) drawn out by participant E1 and digitally recreated.

E1

For E1 (Figure 49), receiving the invitation email was considered an anxious part. Before reading about the contents of the email, she was worried about why she is receiving this health-related email. By reading through the invitation email, she became more calm about the service. The process of booking an appointment was considered calm, but the participant described herself as cautious about the service. However, she was confused about the meaning of "book an appointment", since the prototype included a similar phrasing for booking an appointment for the blood test as well as booking an appointment for a follow-up consultation with a specialist.

Once she received her high PRS result, she reported becoming much more anxious, for which the following information, including the metaphor, calmed her down. After becoming aware of the fire safety metaphor, she reported her emotional state as stable, yet cautious about the next steps. She mentioned the importance of scheduling an appointment for breast cancer

screening as soon as possible, indicating that she plans to act promptly to address the high genetic risk. E1 further mentioned that she would pay attention to the lifestyle recommendations and consider integrating them into her daily life.
Despite the high-risk result, she mentions that the clear communication and disclaimers help her to stay calm. She acknowledged the importance of the result but did not panic, understanding that the result does not pose an immediate threat.

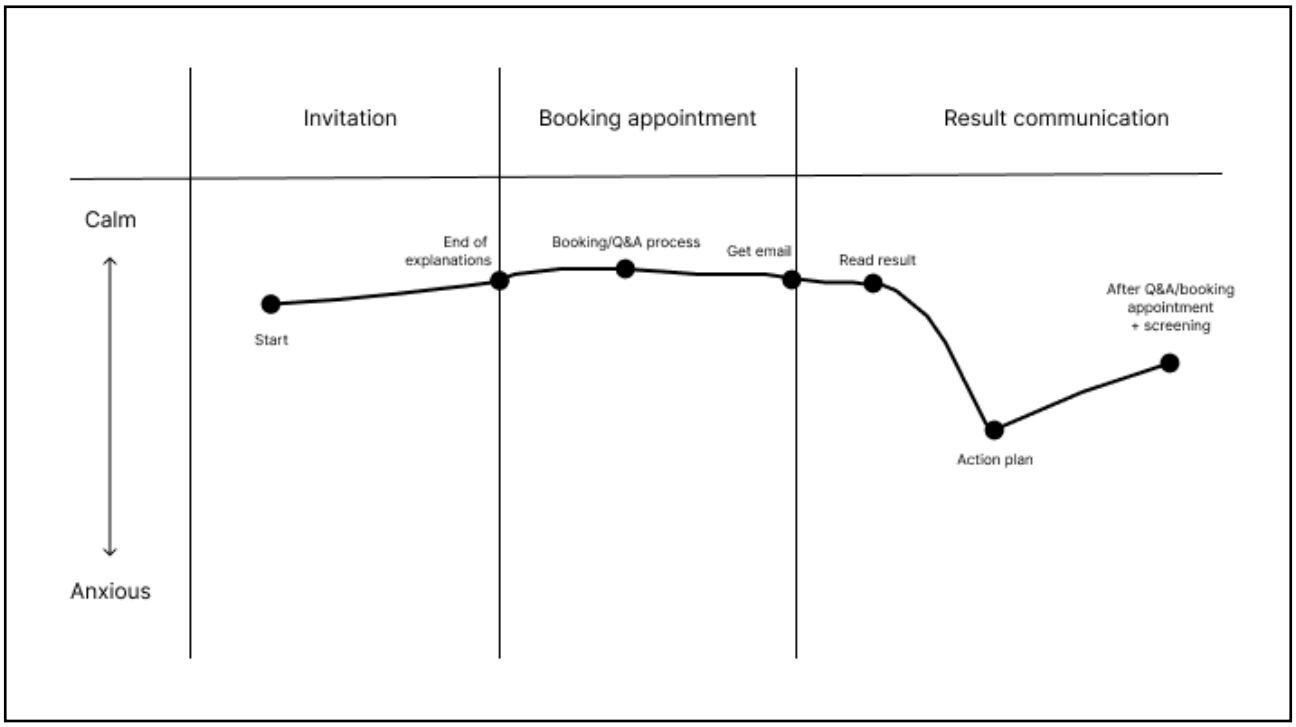


Figure 50: The Emotional Journey (Open Practice Library, 2022) drawn out by participant E2 and digitally recreated.

E2

Throughout the initial email invitation and the blood test appointment booking stage, E2 (Figure 50) described her emotional state as relatively calm. However, after reading the high-risk result, she described herself as more anxious. After reading through the "Action Plan" part on the result communication page, she considered herself increasingly calm. She found the use of the fire safety metaphor and visuals particularly calming. She mentioned that it was nice to have calming visuals and metaphors, as these elements made the information more relatable and less formal, which can often feel formal in official services. She was keen to click on the "Read more" buttons throughout her user journey, highlighting that she values detailed descriptions. After participating in the service, she would consider herself to talk with family and friends and schedule a screening.

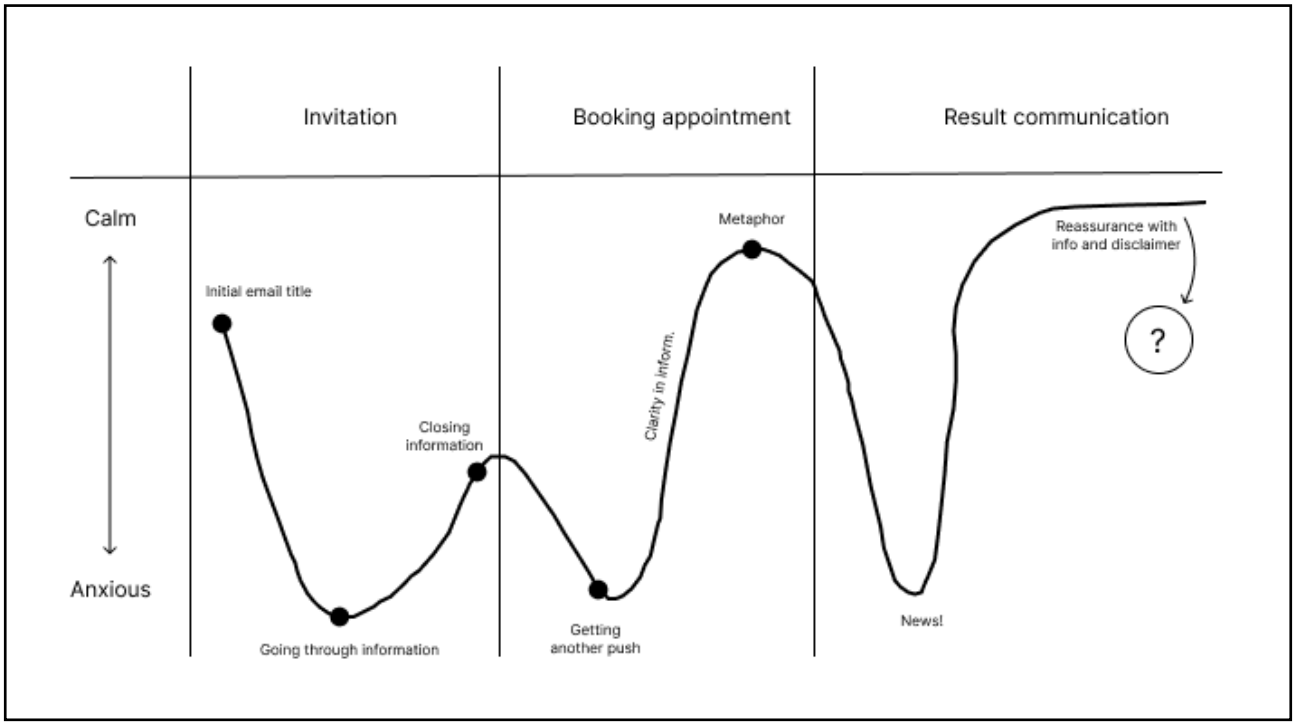


Figure 51: The Emotional Journey (Open Practice Library, 2022) drawn out by participant E3 and digitally recreated.

E3

Among the four participants, E3's highlighted Emotional Journey (Figure 51) had the most fluctuations throughout the evaluation. When receiving the email, she was unsure why she is receiving this email and thus worried whether it means that something is wrong with her health. She considered the amount of information in the invitation email overwhelming, causing more anxiety. After reading through the invitation email, she is somewhat calmed down, but still rather anxious. This reminder highlights again her initial concerns and fears about the health implications of the service. However, by this point the information becomes more clear to her, aided by the fire safety metaphor. After the fire safety metaphor, she describes herself as roughly as calm as at the beginning of the evaluation.
In the next step, she gets to know that the test result is high, which she highlights with an anxious state of mind. However, she is calmed down after reading through the information on the result communication sub-pages. She considers herself reassured by the information provided, especially the disclaimers. However, there is still an underlying concern about "What's next?", relating to confusion about the follow-up steps. She noted that since she is not expected into the screening until the following year, she would expect to talk to family and friends about the follow-up steps.

E4

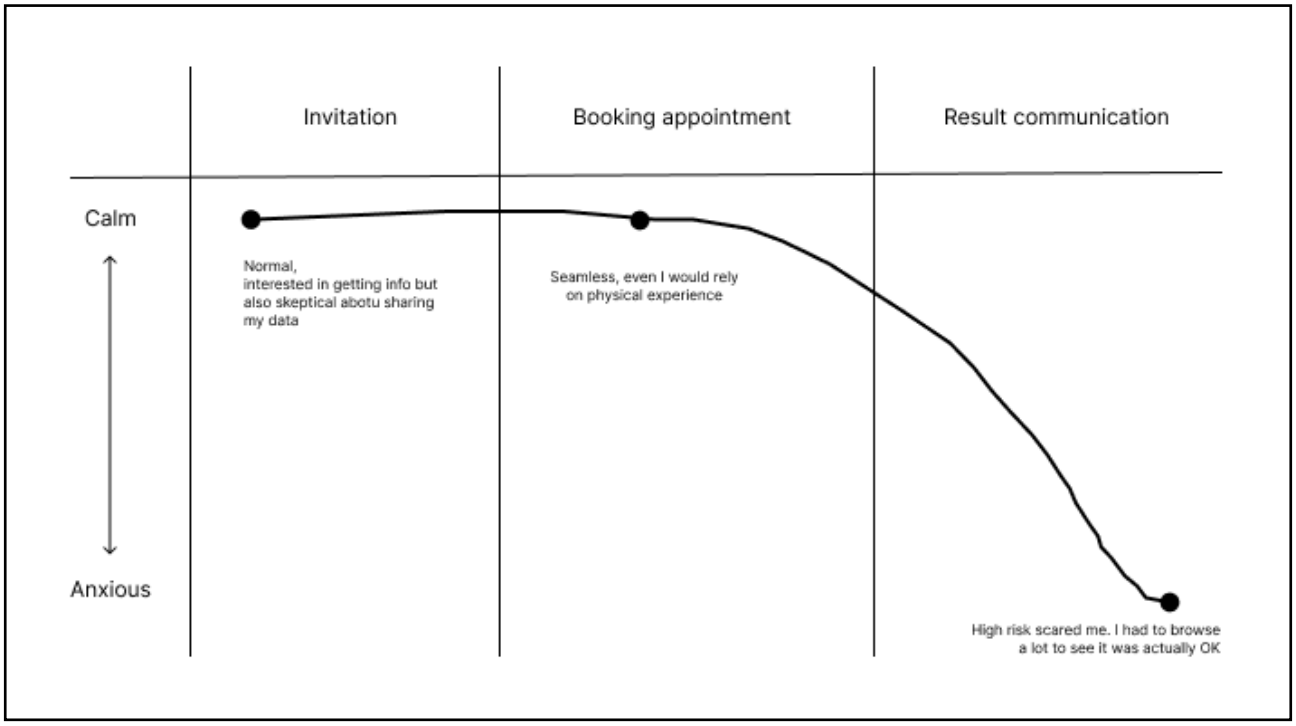


Figure 52: The Emotional Journey (Open Practice Library, 2022) drawn out by participant E4 and digitally recreated.

Among the participants, E4's (Figure 52) emotional state was the most anxious as a result of the service. During the first two stages, she considered her emotional state relatively calm. In the invitation stage, she was interested in getting information about the service but was also very skeptical about how her data is going to be used. She considered the appointment booking process seamless but highlighted that her actual experience of the service would be determined in significant part by the interaction with the healthcare professional taking the blood test. In the result communication stage, she noted that the high risk scared her and that she had to browse a lot of the information on the sub-pages to understand that her situation was not the actual situation was not as serious as previously feared. The user feels somewhat comforted by the constant presence of support options, such as the banner indicating the availability of specialists for further questions. She highlighted that she would reach out to a healthcare professional to seek reassurance and emotional support. She feels that the current information fails to provide the necessary comfort and direction about dealing with her result.

5.1.6 Discussion and conclusion

Relation to personas of the evaluation participants

This section aims to highlight which kinds of personas (from Chapter 4.1) were represented in the light evaluation sessions. E1 showed a keen interest for detailed information about the service by looking into details via the "Read more" buttons. Her detail-oriented, yet less

anxious emotional state (compared to the Anxious Receiver) suggests that she relates to the Technologically Savvy persona.
E2 discusses the importance of understanding the implications of the test result without panicking, appreciating clear and concise information, but also the opportunity to read into more detail about the result using online resources, such as Google. She focuses on taking practical steps to manage her health. Her practical mindset relates to the Pragmatic persona, however, her willingness to read in more detail also relates to the Technologically Savvy persona.
E3 demonstrates a clear, practical approach to health information. This participant values actionable information and efficiency in communication, which aligns with the Pragmatic persona's traits of focusing on the next steps and understanding practical implications.
E4 aims to get in touch with a healthcare professional after receiving the high-risk result. She was generally anxious about receiving the score, thus she likely relates to the Anxious Receiver persona.
In conclusion, three of the four personas were at least partly observed among the participants. The Indifferent persona was not observed. From the original user study, the three personas represent 11 (out of 12) user interviews.

Understanding the concept of genetic risk and the role of My Genetic Mirror

The evaluation sessions revealed that the users grasped the basic concept of genetic risk and the role of the My Genetic Mirror service in assessing and managing this risk. Participants understood that genetic risk is inherited and can influence the likelihood of developing certain conditions.

Emotional responses to the provided information

Users exhibited a range of emotional responses throughout their interaction with the service. The initial invitation email was considered by some to be overwhelming and that there should be an at-a-glance overview about the information. It was noted that although the process of booking an appointment for the blood test was streamlined, the actual experience would depend on the in-person interaction with the healthcare professional taking the blood test. Multiple users felt anxious or confused, especially when encountering terms like "high risk". However, when provided with more detailed information, and especially when good news was included alongside the bad, users reported feeling more at ease and supported. The prototype mentions that the user is assigned to a screening program from the next year, however, this is considered to be too far in the future.

Likelihood of taking action based on the information provided

Participants indicated a strong likelihood of taking recommended actions based on the information provided, such as scheduling follow-up screenings or making lifestyle changes. Users valued clear and actionable steps, and the option to consult with specialists was seen as essential for encouraging follow-through. Personalized recommendations that considered individual medical histories and family health backgrounds were considered more motivating than generic advice. However, any confusion or perceived lack of immediate applicability could diminish the likelihood of users taking action. To enhance follow-through, clear timelines, step-by-step guides, and reminders are recommended.

Ease of navigation through the online demo

The overall navigation of the online demo was reported to be intuitive, with a clear progression from one section to the next. However, some areas required improvement to prevent user disorientation. In the invitation email, it was noted that the information can be overwhelming for the person and that a better overview of the parts would be necessary. On the other hand, the result communication sub-pages with their navigation bar menu were perceived as easier to grasp. The availability of help and support options was appreciated, but users suggested these should be more prominently accessible throughout the navigation process to assist when needed without disrupting their flow.

Most and least calming elements of the service concept

The evaluation identified several calming and anxiety-inducing elements within the service concept. The most calming elements included balanced information that presented both positive and negative genetic insights, helping to create a more balanced perspective and reduce anxiety. Visual aids and metaphors, such as the fire safety metaphor along with the candle element, were considered effective in making complex information more understandable and less intimidating. The availability of support options and easy access to specialists also reassured users, making them feel supported throughout their experience. Conversely, the least calming element was the result with the words "high risk", which induced anxiety. It was noted that the report was somewhat confusing and contradictory since it mentions that the risk is "high" but the chance of getting breast cancer from this is "low". Additionally, it was mentioned that more emphasis should be put into presenting the full result report since it was perceived as calming after getting to know the high-risk score.

5.1.7 Suggestions for improving the design concept

To improve the service design concept, several key suggestions emerged from the evaluation. First, the invitation email should be simplified and clarified to avoid overwhelming recipients,

with an at-a-glance overview of the information provided to make it less intimidating. The booking process should distinguish clearly between booking an appointment for a blood test and a follow-up consultation to avoid confusion, using simplified terminology.
Secondly, in terms of result communication, the "high risk" wording caused significant anxiety for the participants, especially since it was contradictory to the part in the report that highlighted that the chance of getting breast cancer is low. Thus, an alternative phrasing could be considered that highlights why it is important for the person to know the polygenic risk score but that simultaneously doesn't phrase the risk as "high". However, it should be ensured that the phrasing is medically accurate.
Thirdly, the communication should highlight data privacy and how the collected data was used. A lack of privacy-related information caused anxiety about taking part in the service (E3 and E4). Fourth, it should be made more clear why the person is invited to the service. Participants expressed worry about why they are included in the service. Although the information was mentioned in the invitation email, it was highlighted that this information should be communicated as early as possible in the email.
Fifth, the lifestyle recommendations can be made more tailored to the person's health by incorporating further health data. Sixth, the fire safety metaphor module should have been included earlier, especially on the result communication page. Although it was mentioned that the fire safety metaphor was a significant factor in helping the participant make sense of the result and calming the person down, it was highlighted that the metaphor should be mentioned as the first thing in the result communication. By including the metaphor earlier, the person would have an introduction to how to think about the result even before receiving the high-risk score.

Conclusion of the light evaluation

The light evaluation of the My Genetic Mirror service concept provided valuable insights into user perceptions and usability of the interactive prototype. Participants grasped the concept of genetic risk and appreciated the detailed, clear communication, though they found some elements anxiety-inducing and confusing, particularly the term "high risk." The evaluation highlighted the importance of balanced information, visual aids, and accessible support to mitigate user anxiety. Clear, actionable recommendations were valued, yet the confusion over terminology and data privacy concerns indicated areas needing improvement. To enhance the service, simplifying the invitation email, clarifying appointment booking processes, improving result communication, and emphasizing data privacy and the rationale for participation are recommended. These refinements can help ensure a more supportive and comprehensible user experience.

5.2 Contribution to new knowledge

This thesis provides significant contributions to the field of digital health communication, particularly in the context of communicating PRS for BC. The following highlights several key areas where new insights have been gained throughout this thesis.

User-centric design in health communication tools

The study reveals the importance of user-centric design in developing digital health communication tools. By incorporating feedback from both users and experts, the research highlights the necessity of intuitive visual aids, clear language, and avoiding medical jargon to reduce user anxiety and improve comprehension.

Hybrid communication models

The findings from the user study and the evaluation suggest a hybrid model of PRS communication that combines digital tools with in-person or video consultations, especially for conveying high-risk results. This approach not only leverages the accessibility of digital communication but also ensures the emotional and informational support provided by human interactions.

Educational content and mental models

The need for comprehensive educational content to help users build a mental model of PRS and its implications is another significant insight. Interactive tutorials, explanatory videos, and real-life examples can aid users in understanding their genetic risks and managing their health proactively.

Expert interviews highlight different ways in which genetic counselors support PRS recipients by helping to create a mental model about genetic risk. This includes framing PRS as a small change, highlighting the value of PRS as an early warning, and giving the person a sense of control in managing their risk.

Trust and transparency

Trust in the data handling process and transparency about how user data is used and protected emerged as crucial factors. This emerged from the user study and the evaluation. Building a reputable brand through partnerships with recognized healthcare institutions can enhance the trust and acceptance of digital health tools. Furthermore, PRS recipients should understand how data privacy is ensured and how their data is handled.

5.3 Limitations, recommendations, and comments

5.3.1 Limitations of the user and expert studies

Sample size and diversity

The user and expert interviews provided valuable insights, but the sample size and diversity were limited. The study included Estonian-speaking women aged about 40 in Estonia, which does not account for individuals who do not speak Estonian.

Potential bias in sampling

The recruitment of participants through network sampling (personal referrals) could introduce selection bias. Participants referred through the researcher's network might share similar characteristics or perspectives, which could skew the results and limit the variability of responses.

Limited inclusion of mock-up reports

Due to time constraints, two interview participants did not discuss the mock-up reports of high and low PRS risk results. This omission might have led to incomplete data regarding participants' reactions to these specific scenarios, potentially overlooking insights on how users perceive and interpret PRS results.

Language and translation issues

The interviews were conducted in Estonian and then translated into English. Despite efforts to ensure accuracy, the translation process might have introduced subtle changes in meaning or missed nuances, affecting the interpretation of the interview data.

5.3.2 Limitations of the design concept and recommendations

Limited focus of conceptualization

The literature review along with user and interview studies highlighted rich insights into PRS communication. However, to make the conceptualization phase manageable given the time constraints of the graduation project, the focus was placed on creating on a mental model of PRS for recipients. This means that the resulting My Genetic Mirror concept does not fully incorporate every relevant finding, e.g., including users' wish to understand data processing and measures for data privacy. However, the proposed concept provides a first prototype for the proposed PRS communication service by EHIF, where the research and conceptualization insights can be used for the real-life service proposition.

Limited inclusivity of the service concept

The following highlights three instances where the service concept has limited inclusivity for different demographic groups. First, although the target group of the conceptualization is broad – 40-year-old Estonian women – it doesn't consider how disadvantaged groups may interact with the service, such as the visually impaired. Secondly, the service concept assumes that the users have Estonian heritage, which is a technical prerequisite for the Estonian Biobank for determining an accurate PRS result. However, certain demographic groups in Estonia do not have Estonian heritage and for them, the PRS result may have little prediction power. Thus, it is important to highlight this limitation in the actual service. Thirdly, genetic BC risk can also affect further target groups, including men (Konduri et al., 2020). Thus, taking into account further target groups of BC risk can enhance the service desirability.

Limited scope of evaluation

A light evaluation of the design concept was performed due to time constraints. This limited evaluation focused on gathering initial feedback and did not allow for a comprehensive assessment of the tool's usability and effectiveness across diverse user groups. Further activities can include a more thorough evaluation of the service concept with participants who are from the exact target group. It can be also interesting to perform a quantitative evaluation to extract further insights into the desirability of the prototype.

Further mediums and channels

This graduation project proposes the service concept with a website and email invitation format. However, further research and creative exploration into different mediums can yield additional insights about possible PRS communication. These can, e.g., include video materials (as mentioned in the user study).

Further target groups

This graduation project focuses on communicating BC PRS to women. However, genetic risk is present in every human. Thus, further activities, as also included in the roadmaps, can relate to tailoring general PRS communication to, e.g., men based on their needs and wishes. Such findings can help to make PRS communication more inclusive by considering further target groups.

5.3.3 Use of AI tools in the context of this graduation project

Throughout the process of working on this thesis, in addition to manual work, different AI tools were utilized. This included ChatGPT 4 in searching for relevant information, helping

improve the readability of the text, generate ideas and help evaluate during the analysis and design process. For finding relevant academic publications, the Consensus.app was used. Furthermore, the free version of Grammarly was used for checking grammar.

5.4 Personal reflection

This Master's thesis represents the most substantial design project I have undertaken to date. With a background in business and engineering from my Bachelor's studies, this Master's program has introduced me to collaborative work in design. My thesis has provided a focused exploration of PRS, an area I find particularly intriguing. This project has afforded me the opportunity to work more independently than ever before, enhancing my knowledge of PRS and significantly improving my interviewing skills.

The rigorous nature of this project required adapting to the challenges of interviewing and understanding a diverse target group with varied life stages, goals, perspectives, and backgrounds. Developing empathy and managing emotional distress in interviewees, especially when discussing sensitive topics, were critical skills I honed during this process. Conducting my thesis research internationally—academically based in the Netherlands while performing fieldwork in Estonia—enabled me to navigate the complexities of Human Research Ethics. This experience highlighted the potential friction points between design processes and the stringent requirements of healthcare domains. As a result, I now possess a greater capability to approach such situations with confidence and experience.

The analytical aspect of the project, which involved processing and interpreting extensive interview data, underscored the necessity of iterative analysis to derive concise, yet rich conclusions. Working independently on the conceptualization stage of the project, as opposed to the team-based approach typically employed during the Master's program, was a novel experience. Identifying whether a design proposal effectively addressed a problem was challenging at times, but feedback sessions with supervisors and discussions with peers and family were instrumental in clarifying my progress.

Overall, this topic, though complex, proved to be immensely rewarding. The process highlighted the development of my research and design skills and offered a valuable opportunity to step outside my comfort zone. Experimenting, facing setbacks, and continuously improving were integral to this constant learning process. The comprehensive and detailed guidance provided by my supervisors was indispensable to this project. I will take the learnings from this project and I am looking to apply them in the healthcare sector in the future.

5.5 Summary

Project context

In light of technological advancements in genetic testing, it is possible to accurately determine an individual's genetic risk for different disease types, including breast cancer (BC; Lewis & Vassos, 2020). To enable the Estonian healthcare system to discover BC earlier, EHIF plans to introduce a personalized BC screening program that utilizes genetic testing to identify an individual's genetic predisposition to BC. The target group of the personalized BC screening program is 40-year-old women in Estonia. Individuals with a high PRS result are allocated into the official BC screening program from the age 41, whereas low PRS individuals are invited to start participating from the current entry age of 50. As part of this personalized BC screening program, EHIF aims to communicate the PRS result to the participants after genetic analysis. (estPerMed I, 2021) However, since PRS is a new concept to the general population, EHIF expects the recipients to react to the result with worry, possibly seeking consultations from a healthcare professional and thus putting further strain on the healthcare system. Since PRS does not pose an immediate threat to the person's health, no explicit consultations are intended for the service. Thus, EHIF aims to communicate PRS results to recipients by providing the necessary support via a digital tool. This Master's thesis focused on conducting qualitative research into the support needs of women receiving a BC PRS and designing a digital communication service to create peace of mind for PRS recipients after receiving the test result. The resulting findings and the design concept are intended to be presented to the Estonian Health Insurance Fund (EHIF) as input for the design of the digital PRS communication service.

Literature review

To analyze existing research into PRS communication, a literature review was conducted. There, breast cancer, PRS, and the relation between the two was explored, along with existing services from the private sector, and informational and social support needs for women receiving a BC PRS. As a result of the literature review, three areas were highlighted as requiring further research. These included uncertainty about the extent to which BC PRS communication can be performed digitally, how a digital tool should look like that provides the necessary information to the PRS recipient without needing to see a healthcare professional for a consultation, and how to ensure that the digital communication tool provides adequate social support.

Interview studies

To understand these aspects better in the context of the target group, 12 user interviews (about 40-year-old women in Estonia) and 3 expert interviews (genetics experts with a background in

BC PRS communication to recipients) were conducted. The interviews were semi-structured and interview guides were used. The interviews were recorded, transcribed, and translated from Estonian to English. Thematic analysis (Braun et al., 2023) was used to formulate themes based on the interviews. From the user interviews, context factors and five themes were identified. The five themes include overwhelming information during PRS communication, recipients overstating the concept of genetic risk, recipients looking for clarification about the implications and practicalities of the PRS result, and their stance on being communicated PRS online. The expert interviews yielded three themes. They relate to experts suggesting different approaches to phrasing the polygenic risk to calm down recipients, highlighting the methods that genetic counselors use to create a mental model about PRS for the recipients, and ethical concerns about PRS communication.

Conceptualization

The design goal for the conceptualization phase was to reduce the proportion of women reaching out to a medical professional after engaging with the digital PRS communication service. First, the findings from the interview studies were revisited by employing a persona technique to gain a more thorough understanding of the different user types. The technique yielded four distinct personas: the Anxious Receiver, the Technologically Savvy, the Pragmatic, and the Indifferent. These results highlighted that although all of the four personas are open to initial PRS communication via a digital channel, some users (relating to the Anxious Receiver persona) wish for an in-person consultation. To clarify the requirements for the design concept, design criteria were formulated based on the literature review and interview studies. As the literature review and the expert study had provided insights into the techniques genetic counselors use for creating a mental model for users to understand and relate to the concept of genetic risk, a How Might We statement was formulated as: "How might we use genetic counseling techniques to create peace of mind for female recipients of a high PRS result?" Since receiving a hypothetical high PRS result was related to more worry, anxiety, and stress, the conceptualization stage focused on communicating high PRS. During the conceptualization phase, different design methods were used, including mind-mapping (Van Boeijen et al., 2014), a modified ViP method (from IDE Master's course "Context and Conceptualization"), a brand ladder & a category migration (Wansink, 2003; In combination with the format from the "Brand and Product Commercialization" course at IDE), and a Creative Facilitation Session (Heijne & van der Meer, 2019). These methods served in creating the service concept called My Genetic Mirror, a sub-page on the Estonian Health Portal platform. The concept consists of four key parts: changing the perception of the service towards an identity focus, simplifying the concept of genetic risk and the recipient's relation to it with a fire safety metaphor, spreading out information delivery over a longer time period

to avoid overwhelming information, and providing a Follow-Up Questions module for follow-up questions. A service blueprint and a digital prototype were designed, which serve as input for potential immediate implementation by EHIF.
To consider how the service concept could look like in the future, a strategic and a tactical roadmap were designed. For this, creative trend research, the creation of a future vision for the roadmap, a time-pacing strategy, and technology scouting (Simonse, 2018) was done. The future vision for 2030 was formulated as creative emotionally reassuring yet cost-efficient genetic health management for peace of mind. The first horizon focuses on raising awareness about genetic risk by providing the first insights into BC PRS. The second horizon focuses on enhancing the accessibility and comprehensibility of genetic risk assessments. The third horizon focuses on helping the person stay on top of managing their genomic risk via an AI assistant.

Light evaluation and conclusion
To evaluate the service concept, a light evaluation (Van Boeijen et al., 2014) was performed with four students from IDE. Apart from one participant showing characteristics of the Anxious Receiver personality, the prototype was deemed helpful in understanding a hypothetical high BC PRS result by the other participants. They highlighted the visual aids, the use of the fire safety metaphor, and the balancing of both positive and negative genetic insights. However, the prototype was met with criticism in causing anxiety, worry, and confusion, especially the wording "high risk" in the result, and an overwhelming amount of information in the invitation email.
The thesis concludes by highlighting the contribution to new knowledge, limitations, recommendations, comments, and a personal reflection on the process.

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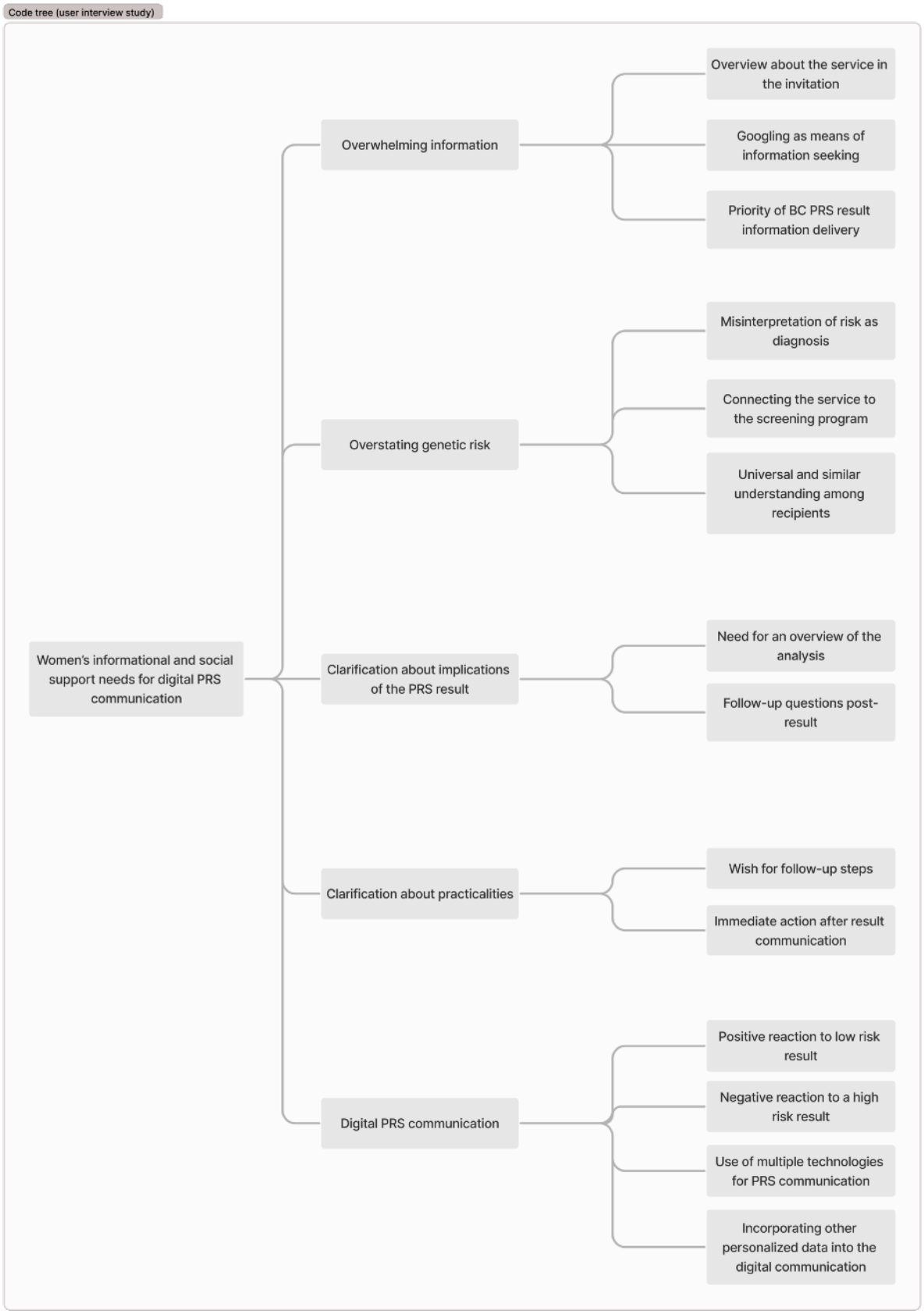
Throughout this thesis, the course material from the following Master's courses at the faculty Industrial Design Engineering of TU Delft were used in particular: "Design roadmapping", "Capita Selecta Medisign", "SPD Research", "Brand and Product Commercialization", "Health Systems Transformation", and "Context and conceptualization". The graduation project report and deliverables include images from Flaticon.com, Icons8.com, and Storyset.com.

Appendix

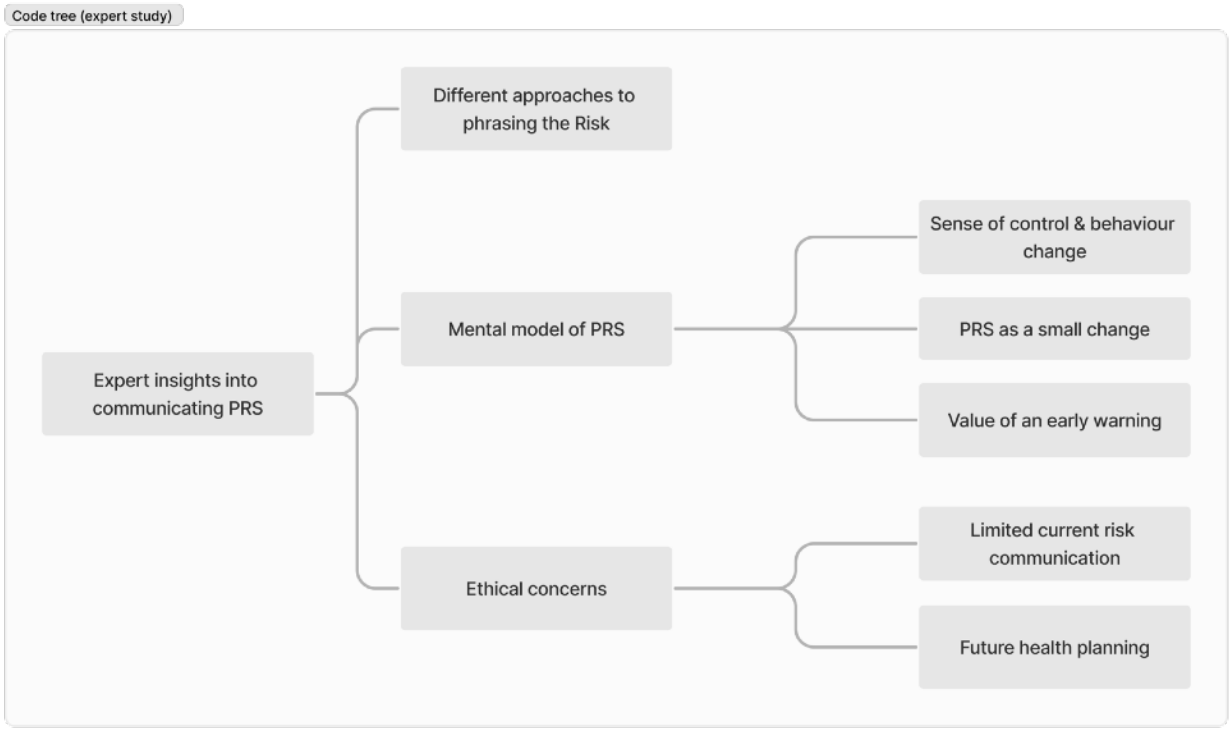
Appendix A

The resulting code tree from the user interviews after thematic analysis.



Appendix B

The resulting code tree from the expert interviews after thematic analysis.



Appendix C

Interview guide used for the user interviews.

Interview with 40-year-old women

Sample: 10-15 women between 38-42, maximum variability sample: urban, rural, low education, high education, ...

Research question: What informational and social support do women receiving a polygenic risk score for BC need in the context of a digital communication tool?

0. Introduction (5 min)
Introduction of the interview, getting consent from the participant and mention of the use of audio recordings.
For me: Briefly introduce yourself and mention the objectives of the interview. Mention that the interview will last 45 minutes and that I expect to cover a number of topics. Explain how the interviewee's input will contribute to the research and service development.

I. Background and general questions (15 min)
1. Please tell me a little about yourself (age, profession, optional (respondent does not have to reply): what has been your exposure to cancer? (family, acquaintances, friends, etc.)
2. If you had to explain breast cancer as such to a 12 year old child, how would you do it? If you had to explain what increases the risk of breast cancer, how would you do it?
3. What have you heard about the breast cancer screening program offered by the Estonian Health Insurance Fund? Could you explain in simple words what the program is about?

II. Personalised breast cancer results report (15 min)
4. If you have had a health problem, how have you sought information about it? Who do you talk to?

Imagine that, in a few months, you receive an email inviting you to take part in a personalised breast cancer screening programme, offering you the chance to have a genetic test to assess your genetic risk of breast cancer. If you are already a gene donor, the hospital will be able to use the data in the Biobank for the service. You will be shown the result of the analysis in the genetic counsellor's office.

5. How would you react to such an e-mail? What thoughts and feelings would it evoke?

6. If you had to describe to someone in simple words what a risk score is, based on your current impression, how would you do it?
7. What would you expect from a meeting with a genetic counsellor? How would you feel before the visit?
8. What information would you expect to receive BEFORE the results of the breast cancer risk score? (e.g. description of disease)

But now imagine that breast cancer risk score counselling is replaced by a digital service. You access the results digitally.
9. What general information would you expect from this digital service?
10. How could this service talk about the risk (and the situation) in a meaningful way?
11. How would you feel before accessing the results?
12. In this case, what is important to you when you read the results?

Let's assume that the risk score in the risk score report turns out to be low/high (change the variant for each participant).
13. What would you like to experience after learning about the risk score? If support, then what kind?
14. What do you think you would do if the risk score turns out to be low? (what would you need?)
15. What do you think you would feel and do if the risk score turned out to be high?
16. How important is individual counselling or a recommended action plan to manage risk for you?

Let's now imagine that the risk score turns out to be high/low (change the option for each participant).
17. Would this risk score change your attitude towards breast cancer risk? If so, how? Do you think it would change your behaviour in any way?

III. Example of a specific report (20 min)
Give the interviewee the report and have her read it.

18. What are your first impressions on seeing the report?
19. How would you describe the comprehensibility of the information presented in this report?
20. What do you find particularly positive or particularly negative?
21. What could this advice be? From whom? How long? What should the counsellor talk about?

Let the participant describe.

22. Do you think that the genetic counselling service you describe could be replaced by a digital service (as shown in this report)? Why? Which parts could be, which parts not?
23. Do you think the report could use more information or features? If yes, which ones?

IV. Final questions and conclusion

24. Do you have any recommendations or preferences for a digital service? Do you have any questions for me or about the project?

Thank the interviewee for her time and contribution. Mention the next research steps. Provide contact details in case the participant has any further questions or they would like to be kept up to date with the progress of the research.

Appendix D

Interview guide used for the expert interviews.

Interview with experts

Sample: 3-5 genomic experts and genetic counsellors.

Research question: What informational and social support do women receiving a polygenic risk score for BC need in the context of a digital communication tool?

0. Introduction (5 min)

Introduction, getting consent from the participant and mention of the use of audio recordings.

Me: Briefly introduce yourself and mention the objectives of the interview. Emphasise that the interview will last 45 minutes and that I expect to cover a number of topics. Explain how the interviewee's input will contribute to the research and service development.

I. Background and experience of the expert (10 min)

1. Please describe your current job role and experience with genome-based health care, particularly in relation to breast cancer/polygenic risk assessment for various diseases?
2. What do you know about the personalised screening programme? Could you summarize in simple words what you currently see it is or would be?
3. How do you feel about the (breast cancer) personalised screening programme? What opportunities and threats do you see in implementing this programme?
4. In general, how do women react to getting a breast cancer risk score? How are they currently counselled? What works and what more would patients actually want?
5. How do you explain/explain the nature of the breast cancer risk category to those being counselled?
6. If, then what problems do you encounter when giving these explanations?

II. Information and social support (10 min)

7. What is the general role of the genetic counsellor in assisting the counselee?
8. What information does the counselee need after receiving a polygenic breast cancer risk score?
9. What social support does the counsellor need after receiving a polygenic breast cancer risk score? (from genetic counsellor, acquaintances, family, etc.)

10. What are some situations where counselling, or lack of it, can go wrong?

III. Feedback on the digital design concept (15 min)
Imagine that, in a few months, a 40-year-old woman receives an invitation by email to participate in a personalised breast cancer screening programme, where she is given a genetic test to assess her genetic risk of breast cancer. If the person is already a gene donor, the hospital will be able to use the data in the Estonian Biobank for the service. In this case, breast cancer risk in-person counselling has been replaced by a digital service. The woman can access the results digitally.

11. If we imagine that part of genetic counselling could be replaced by a digital service, which parts could be replaced and which parts perhaps could not?
12. What would the digital service need to include to effectively communicate the risk score? How should participants in a screening programme be told what to do with the information they receive?
13. How can it be ensured that the results of the analysis are presented in a way that is understandable to those being counselled?
14. What role could digital tools play in providing emotional support to the counselee?

Present the report. Give the mock-up report to the interviewee and ask him/her to read it.

15. What are your first impressions on seeing the report?
16. How would you describe the comprehensibility of the information presented in this report?
17. What do you find particularly positive or particularly negative?
18. What could this consultation be like? From whom? How long? What should the adviser talk about?

IV. Final questions and conclusion (5 min)
19. Do you have any other suggestions for the development of the proposed digital service concept?

Thank the interviewee for his/her time and contribution. Provide contact details in case the participant has any further questions or they would like to be kept up to date with the progress of the research.

Appendix E

The project brief.

DESIGN FOR our future

TU Delft

Personal Project Brief – IDE Master Graduation Project

Name student Mihkel Annus **Student number** 5850606

PROJECT TITLE, INTRODUCTION, PROBLEM DEFINITION and ASSIGNMENT
Complete all fields, keep information clear, specific and concise

Project title Supporting women receiving a breast cancer risk score in a genomics-based personalized healthcare service

Please state the title of your graduation project (above). Keep the title compact and simple. Do not use abbreviations. The remainder of this document allows you to define and clarify your graduation project.

Introduction

Describe the context of your project here; What is the domain in which your project takes place? Who are the main stakeholders and what interests are at stake? Describe the opportunities (and limitations) in this domain to better serve the stakeholder interests. (max 250 words)

Worldwide, over 2 million individuals get diagnosed with breast cancer annually. For over 600,000 of such patients, the disease is terminal. (WHO, 2023) In Estonia, breast cancer is the most common type of cancer among women, with 19% of total cancer incidence in 2018 (National Institute for Health Development, 2021).
Early detection significantly improves breast cancer survivability (WHO, 2023). Thus, the public Estonian Health Insurance Fund (EHIF) runs a screening program where 50–70 and 74 year old women are invited to a mammography test once every two years.
However, with advances in genomics-based healthcare, it is now possible to estimate a person's risk for breast cancer. This development promises better predictability, better patient cancer survivability, and a more cost-effective value proposition. By providing a blood test and conducting a subsequent laboratory analysis, a risk score can be calculated.
Studies have shown that the risk of breast cancer is already significant before the age of 50, the current lower bound. The EHIF plans to already include 40 year old women to calculate their risk scores. The goal is to onboard the women with a high risk score already at age 41. EHIF is planning to introduce the service in early 2025 for 40 year old female participants. The development of such a service provided by a public institution is currently unique in the world. My role will be to create a service to reduce stress, provide self-management guidance in understanding how to react to a risk score. The goal is to react to participant needs and reduce strain on the healthcare system.

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DESIGN FOR our future

TUDelft

Personal Project Brief – IDE Master Graduation Project

Problem Definition

What problem do you want to solve in the context described in the introduction, and within the available time frame of 100 working days? (= Master Graduation Project of 30 EC). What opportunities do you see to create added value for the described stakeholders? Substantiate your choice.
(max 200 words)

After requesting a risk score for breast cancer, the person is provided with one after they have consented to giving their genomic data through a blood sample. However, at this point the participant has little information about the next steps. Especially in case of a high risk score, the individual may face uncertainty and anxiety about their situation and seek information directly from their GP. But this increases the load on the first line of the healthcare system (GPs). At the same time, only a fraction of the GPs are trained to assist in genomics-based healthcare. Thus, EHIF is seeking a digital design concept to provide participants the necessary information to support them in understanding their risk score and which activities they are expected to do. The goal is reduce the pressure on the healthcare system, as well as inform the women receiving the score to encourage self-management.

Value added for different stakeholders:
- Participants: Higher awareness about their medical condition
- The healthcare system in general: Decreased workload pressure from participants seeking advice on their health condition and risk score
- EHIF: Contribute to a more streamlined and cost-efficient patient journey
- Other healthcare systems (e.g. Netherlands): Provide suggestions for implementing digital solutions in genomics-based personalized healthcare

Assignment

This is the most important part of the project brief because it will give a clear direction of what you are heading for. Formulate an assignment to yourself regarding what you expect to deliver as result at the end of your project. (1 sentence) As you graduate as an industrial design engineer, your assignment will start with a verb (Design/Investigate/Validate/Create), and you may use the green text format:

Research question: What are the information and social support needs of women who receive a personalized breast cancer risk score? Design assignment: Design a digital, human-centered service concept (incl. user-dependent pathways) and roadmap to address the information and social support related needs for women receiving a breast cancer personalized risk score in the context of genomics-based personalized healthcare.

Then explain your project approach to carrying out your graduation project and what research and design methods you plan to use to generate your design solution (max 150 words)

Link to Gantt chart: https://1drv.ms/x/s!AtSfdzZLhf-1t2mrVmJU5shogrhY?e=H5ph5F
Expected activities:
1. HREC TU Delft application
2. Literature review of existing and related solutions in the field of personalized healthcare, look at the health education that arises through potential new interactions, peer groups
3. User research: 10-15 interviews: women (around 40 years of age) who have voluntarily donated their genomic data to the Biobank, GPs, genomics-focused experts (e.g. from EHIF and the genetic Biobank of the University of Tartu, Estonia), breast cancer experts, other stakeholders assisting women in genomics-related inquiries.
IMPORTANT: As the service does not yet exist, no participants with previous experience with the service can be recruited. Instead, the interview participants will consist of women in Estonia who have voluntarily donated their genomic data to the Biobank, who will be the target group of the service.
4. Conceptualization: Service pathways & roadmapping
5. Validation and feedback on design proposal: testing with about 5 participants (individuals, medical professionals, genomics-focused experts – to be specified)

Project planning and key moments

*To make visible how you plan to spend your time, you must make a planning for the full project. You are advised to use a Gantt chart format to show the different phases of your project, deliverables you have in mind, meetings and in-between deadlines. Keep in mind that all activities should fit within the given run time of 100 working days. Your planning should include a **kick-off meeting**, **mid-term evaluation meeting**, **green light meeting** and **graduation ceremony**. Please indicate periods of part-time activities and/or periods of not spending time on your graduation project, if any (for instance because of holidays or parallel course activities).*

Make sure to attach the full plan to this project brief.
The four key moment dates must be filled in below

Kick off meeting	February 13th, 2024
Mid-term evaluation	April 4th
Green light meeting	June 10th
Graduation ceremony	July 8th, 2024

In exceptional cases (part of) the Graduation Project may need to be scheduled part-time. Indicate here if such applies to your project

Part of project scheduled part-time	
For how many project weeks	
Number of project days per week	

Comments:
Vacation between April 15th and April 19th

Motivation and personal ambitions

Explain why you wish to start this project, what competencies you want to prove or develop (e.g. competencies acquired in your MSc programme, electives, extra-curricular activities or other).

Optionally, describe whether you have some personal learning ambitions which you explicitly want to address in this project, on top of the learning objectives of the Graduation Project itself. You might think of e.g. acquiring in depth knowledge on a specific subject, broadening your competencies or experimenting with a specific tool or methodology. Personal learning ambitions are limited to a maximum number of five.
(200 words max)

I believe that personalized healthcare is a key to improving patient care and preventing disease on different levels. When the Estonian Biobank launched a campaign to gather genomic data from the population, my dad suggested me to sign up because it may help identify a genomics-related disease earlier and improve related care. As the project has been in works for about 6 years since then, I am curious about how the project is progressing, and importantly, to contribute to it.

My Strategic Product Design Master's has introduced me to strategic design thinking methodologies such as roadmaps, as well as healthcare design tools as part of the Medisign specialization. As I am interested in both, I would like to combine those existing skillsets and develop them as a strategic healthcare designer.