Preventive genetic testing

Valorisation

During the research for this thesis, many opportunities for valorisation of the acquired knowledge arose and some were actually seized. This chapter addresses these opportunities and more, and discusses societal relevance of the results of this thesis.

A majority of the work conducted for this thesis was dedicated to developing a reliable, valid, functional and practical personal disease risk testing service. This service, Health Potential, will soon be launched on the international market. We, as the Maastricht University Health Potential team, are based in a valorisation hotspot, with the proximity and opportunities of Brightlands/Maastricht Health Campus, several universities (e.g. Eindhoven, Ghent, Hasselt and Aachen), the EU region Meus-Rhine (Dutch: Euregio Maas-Rijn), and of course Maastricht University's 'Maastricht Valorisation Centre', providing an excellent accelerator for diffusion and diversification.

I will first discuss the relevance of the results of this thesis for different potential stakeholders. Then, because involvement of stakeholders in product development is essential for successful products, I will conclude with one example of how we involved end-users in development: usability testing of our online Health Potential platform.

Relevance of this work for different stakeholders

The individual

One of the primary and perhaps most obvious target populations is the individual end-user; the customer. These individuals are most likely to benefit from the Health Potential service directly, through acquiring information and starting points for behaviour change. In addition, it is likely fulfill other needs through the 'fun-factor' and satisfaction of curiosity. This may particularly be the case for the early adopters who might generally be more curious about the service in general compared to later adopters who might learn about it when searching for health-related information.

The health care professional

Health Potential is a source of personal health-related information, which can be used for different purposes including supporting health behaviour change. As such, it can be used and implemented in many different health-care settings and different types of health care professionals (HCPs; including dieticians, physiotherapists, weight loss coaches or even nurse practitioners) can use Health Potential concurrent to or merged with their own services. As we cannot expect every HCP stay up-to-date with all disease risk knowledge available, which expands at an astonishing rate, services like Health Potential offer them a means to stay on top of the evidence. Their role is then no longer to collect the information, but rather to help their patient or client to interpret it, place

it in context of their personal situation, help them to translate it to their desired use, and finally support them in the maintenance of their lifestyle change.

In addition, results from chapter 4 provide suggestions as to who might need additional assistance to avoid adverse psychological effects, thereby making the service delivery as safe as possible.

Entrepreneurs and investors

Our convenient location with border-crossing interactions between Maastricht University, Hogeschool Zuyd, and local businesses and start-ups, offers an excellent environment for exploitation of opportunities. This contributes to easy contact with (other) entrepreneurs and investors, resulting in interesting partnerships and collaborations.

Health Potential as a product, as well as its methods, is an excellent basis for diversification, which can lead to new collaborations and initiatives. Currently, different products with a similar approach are being developed, both on our initiative as on request from other parties. These products are based on a similar method as Health Potential but address a different domain. One such example is Health Potential's sister product, Sport Potential, which assesses athletic potential based on one's genetic profile. Similar to the research presented in chapters 3 and 5, for Sport Potential we analysed the genetic mutations associated with different athletic characteristics in a similar manner as for Health Potential.[1]

In addition, chapter 2, which provides information on predictors of interest in DTC-GT, may be useful for this group too as this will help to identify their target population and thereby market their products more effectively and appropriately.

Employers and insurance companies

Both employers and insurance companies have a similar benefit from services as Health Potential: improving the health of their employees/clients to reduce their own costs. Namely, absenteeism and reduced productivity, which are associated with poor lifestyles[2], is a major source of costs for employers. Similarly, health care or disability insurers would have to pay up less if people were healthier. As a result, increasing amounts of attention, time and money are being spent on promoting health of their employees and clients, in which (services like) Health Potential could play a role.

At this moment, first steps have been taken to cooperate with employers and insurance companies. For this, a lean version of Health Potential will be used, including non-

genetic factors only, due to potential privacy and discriminatory issues related to the use of genetic data. Although we are still in the preparatory phases of this collaboration, prospects appear to be good.

Society

Value for society as a whole will primarily be achieved if Health Potential is effective in improving lifestyle and thereby improving future health. A healthier population will have increased production, decreased sick-leave associated costs, and on the long-term reduced health-care costs. However, the impact on society on the short-term is likely to be small, if any, as only a relatively small group of people will initially take up Health Potential and this group is likely to already be more health minded, thereby possibly not addressing the population most at need of lifestyle improvements. In addition, for behaviour change to be maintained optimally on the long term, thereby achieving observable benefits for society, a multidisciplinary approach with interventions at micro, meso and macro levels, is not just desirable but likely a necessity. Finally, for optimal benefits for society, it is a prerequisite that Health Potential does not lead to (unnecessary) downstream medical costs.

Regulatory bodies

Chapter 4 provides input on the balance of pros and cons in the discussion of allowing DTC-GT on the market. As such, it showed that DTC-GT does generally not lead to adverse psychological effects, has moderate impact on behaviour change, and may lead to downstream medical costs. These findings can be taken into consideration during drafting of regulations. In addition, chapter 2 provides input for information provision to the general population regarding DTC-GT. This information should ideally be provided by independent bodies and will enhance informed decision-making of undergoing testing.

Involvement of end-users: the example of our online platform

To enhance acceptance by and value for end-users, we aim to closely involve different stakeholders in development of our products. This sub-chapter describes the involvement of future coaches and clients in the development of our online platform, which is designed to assist both coaches and clients in monitoring the process of testing. Here, both parties can view the current status and respond to required actions. To develop this online application, we collaborated with Hello Sunshine (Heerlen, The Netherlands), and applied a user-centred design cycle, which consisted of multiple short sprints or iterations during which the product was continuously improved: a cyclic process of

design, evaluation and redesign[3,4]. Our goal was to develop a platform that would be convenient and intuitive to use. After several initial cycles based on interaction between Health Potential researchers and developers, potential end-users (clients and coaches) and the application developers, a first version had been developed and was tested and improved in future users, combining different usability testing methods.

User test 1: Heuristic evaluation

Methods

The first version of the system was evaluated using heuristic evaluation, using Nielsen's heuristics (Table 1)[5]. These heuristics are general principles that are common properties of usable systems.

To identify a variety of usability issues[6], 11 evaluators were recruited including usability testing experts, technology experts, and application domain experts. All evaluators had great interest in applications and use of technology and used technology on a regular basis. Before testing, each evaluator was trained in Nielsen's heuristics and was reminded of Health Potential and the purpose of the system. They were asked to navigate through the application at their own pace, doing and clicking as they pleased, writing down their findings, both positive and negative.

All findings were collected and analysed for usability issues (defined as not being able to advance or being distracted or discouraged from a flaw in design or content of the application). At minimum, the critical issues were resolved before we continued with user testing of the next version of the application.

Table 1 Nielsen's 10 heuristics for user-friendly interface design

1. Visibility of system status
2. Match between system and the real world
3. User control and freedom
4. Consistency and standards
5. Error prevention
6. Recognition rather than recall
7. Flexibility and efficiency of use
8. Aesthetic and minimalist design
9. Help users recognise, diagnose, and recover from errors
10. Help and documentation

Results

The heuristic evaluation identified a total of 222 unique issues with the application. Issues

mainly regarded difficulty navigating through the application and the questionnaire in particular. Evaluators mentioned that they missed a quick navigation option for the questionnaire, which then had to be navigated one question at the time, and that some icons for menu buttons were unclear. They suggested the use of a breadcrumb (i.e. a navigational element showing the current page in relation to previous pages visited or a certain hierarchy of pages) for easier orientation in and navigation through the application. In addition, they experienced confusion with validation of answers and skip logic within the questionnaire (e.g. skipping female-only questions for males). In addition, it was considered odd that notifications were not clickable, and ideally they should lead to a location the notification referred to. Finally, errors or ambiguities in textual content were identified, particularly within the questionnaire, and evaluators expressed a wish for additional information at certain locations.

User test 2: eye tracking and 'thinking aloud method'

Methods

We did two series of user testing: one with prospective coaches and one with prospective clients. For this, we used the concurrent think-aloud approach combined with eye tracking. This method involves encouraging the participants to verbalise their thoughts while navigating the application. At the same time, we used eye tracking to allows more objective evaluation of system design and the different interactions of a user with the system which can not easily be observed otherwise[7]. One can evaluate the amount of processing that the user applies through the time that he or she fixates on one design element[8].

Each test was centred around a set of scenarios, describing the context behind why a user would be using the application, including intake, collecting of data and delivery of the results. The scenarios were shown to the participants on the screen to be read and additional instructions were given by the facilitator of the test during the execution of the test.

The sessions were video recorded and transcribed verbatim. These transcripts were then used to generate a list of usability issues, which were rated on severity and assigned the location and interface aspects, like was done during heuristic analysis.

Results

In total, four coaches and five clients participated. All coaches were physiotherapists or exercise scientists. Together they identified a total of 62 unique issues. One menu icon (the hamburger menu icon; figure 1) that had not been changed as it was thought to be a commonly used icon, still caused confusion. However, users quickly figured out what

it meant and how to use it. In addition, the hover-effect, the technique that the design of an element changes when the mouse cursor is on that element and which was thought to give the impression that notifications were clickable, had been removed but this had apparently not resolved the issue. Finally, two coaches mentioned that although the design of the application looked functional, it could use a 'flashy layer'.

Figure 1 Hamburger menu icon

One of the most important areas of the application for the user is the timeline. This shows the user's progress through the entire Health Potential procedure, and is thought to help the users monitor and accelerate their testing process by showing which steps have been completed, what needs to be done or what the user is currently waiting for. Testing showed that all-but-one user interpreted the timeline correctly as multiple processes running parallel to each other (Figure 2) and it seemed to support their perception of the overview of the process.

Continued evaluation

After these two testing rounds, a new version of the application was developed, which was subsequently further improved through multiple informal evaluation rounds with the Health Potential team, development team and other relevant stakeholders. Right before the formal launch of Health Potential, the application, together with all other processes of Health Potential, will be tested as a whole in a pilot run with actual consumers. Naturally, it will remain an on-going process of development, continually improving and expanding the options and possibilities that we can offer. However, this cycle of design, evaluation and redesign nicely illustrates how such cycles could look.

Figure 2 Screenshot of timeline with parallel running processes

In user testing greater focused was placed on the risk report than during heuristic evaluation. Issues in the report were mostly related to interpretation of the information therein. Although some layout changes were suggested that would reduce confusion, such as a change in the order of presentation of genetic and lifestyle factors, most of the issues required additional training, counselling or a more elaborate help function, which are now being developed.

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