TALLINN UNIVERSITY OF TECHNOLOGY
School of Information Technologies

Kristiina Maajärv 153400YVEM

POPULATION AND HEALTH PROFESSIONALS VIEWS ABOUT CARDIOVASCUAL DISEASE PREVENTION FROM THE PERSONALISED MEDICINE POINT OF VIEW IN ESTONIA

Master's thesis

Supervisor: Margus Viigimaa
MD, PhD

Tallinn 2018

TALLINNA TEHNIKAÜLIKOOL
Infotehnoloogia teaduskond

Kristiina Maajärv 153400YVEM

ELANIKKONNA JA TERVISHOIUTÖÖTAJATE SUHTUMINE SÜDAME-VERESOONKONNA HAIGUSTE ENNETAMISESSE PERSONAALMEDITSIINIST LÄHTUVALT

magistritöö

Juhendaja: Margus Viigimaa
MD, PhD

Tallinn 2018

Author’s declaration of originality

I hereby certify that I am the sole author of this thesis. All the used materials, references to the literature and the work of others have been referred to. This thesis has not been presented for examination anywhere else.

Author: Kristiina Maajärv

14.05.2018

Abstract

Background: Cardiovascular diseases (CVD) are the main cause of death in Estonia and worldwide and it has a negative socio-economic impact on the society because mostly, people in the working age develop CVD. CVD are generally highly preventable through healthy lifestyle habits and personal engagement in preventive activities. The key factor of personal CVD prevention is high health literacy and understanding, engagement and attitude towards CVD preventive activities. *Aim*: To assess the acceptance and understanding of the genetic information by population and health professionals in the CVD preventive process. *Method*: Descriptive quantitative research 04.04.2018-30.04.2018. Two questionnaires have been composed, to assess separately the population and health professionals' views and opinions. *Data gathering*: Data was obtained from two sources – data from population and from health professionals' surveys has been collected. *Results*: The results show that population and health professionals would accept the idea of including personal genetic test results into the current CVD preventive activities. Both respondent's groups did favour the information presented to them in a summarized format: 70% of population and 65% of health professionals would favour receiving information in summarized electronic format. Population would slightly more (70%) consider making changes to their lifestyle, when genetics is involved in comparison to when family doctor would be involved (67%). Health professionals see value in having access to patients' genetic data (77%). *Conclusion*: It is evident from the survey results, that population and health professionals would accept the ideas of personalised medicine in preventing CVD and they would like to receive access to the health data in summarized form. There is need for new formats of receiving access to own health data. Therefore, further investigation is needed about the exact format and the way genetic data should be presented to the population as one additional way for strengthening personal motivation, engagement and CVD prevention.

This thesis is written in English and is 62 pages long, including 7 chapters, 18 figures and 1 table.

Annotatsioon

Elanikkonna ja tervishoiutöötajate suhtumine südame-veresoonkonna haiguste ennetamisesse personaalmeditsiinist lähtuvalt

Taust: Südame veresoonkonna haigused (SVH) on Eestis ja mujal maailmas üks peamised surmapõhjuseid ning samuti on sellel negatiivne mõju meie ühiskonnale, kuna enamjaolt noored, tööealised inimesed haigestuvad SVHsse. Võtmefaktor isikliku SVH ennetamistegevuste juures on kõrge tervisekirjaoskus (*health literacy*), tervislikest eluviisidest arusaamine, protsessidesse kaasatus ja suhtumine teemasse. *Eesmärk*: Hinnata elanikkonna ja tervishoiu töötajate suhtumist ja arusaama geeniandmetest SVH ennetamisel. *Meetodid*: Deduktiivne kvantitatiivne uuring 04.04.2018-30.04.2018. Hüpoteesi kontrollimiseks koostati kaks küsimustikku, üks tavainimesele ja teine tervishoiu töötajatele suunatud. *Andmete kogumine*: Andmeid koguti kahest erinevast küsimustikust ja analüüsiti eraldi. *Tulemused*: Nii tavainimesed kui ka tervishoiu töötajad aktsepteerivad mõtet lisada isiklikud geenitestide tulemused praeguse SVH ennetustegevustele lisaks. Mõlemad vastajate grupid pooldasid kokkuvõtet nende andmetest (*patient summary* – arstile ja *health summary* tavainimesele): 70% tavainimestest ja 65% tervishoiu töötajatest pooldasid infot kokkuvõtvas vormis. Tavainimesed kaldusid veidi rohkem oma elustiili muutma, 70% vastanutest, kui geeniandmed olid kaasatud vs 67%, kui perearst seda soovitas. Tervishoiu töötajatest 77% näevad lisaväärtust, kui neil oleks juurdepääs patsiendi geeniandmetele. *Kokkuvõte*: Uuringu tulemused näitavad, et nii tavainimesed kui ka tervishoiu töötajad suhtuvad positiivselt personaalmeditsiini ja sellest lähtuvalt SVH ennetavatesse tegevustesse ning soovivad saada juurdepääsu terviseandmetele kokkuvõttena. Seepärast tuleb edasi uurida, milline on parim formaat kokkuvõte jaoks, milliseid andmeid see peab sisaldama ja kuidas peaks geenitestide tulemusi tavainimesele esitama, et see omakorda aitaks tugevdada inimese isiklikku motivatsiooni ja kaasatust, et aktiivselt panustada SVH ennetamisesse.

Lõputöö on kirjutatud inglise keeles ning sisaldab teksti 62 leheküljel, 7 peatükki, 18 joonist, 1 tabelit.

List of abbreviations and terms

AF	atrial fibrillation
AHA	American Heart Association
BMBF	German Federal Ministry of Education and Research
BP	blood pressure
GRS_{CAD}	genetic risk scores coronary artery disease
CVD	cardiovascular diseases
CHD	coronary heart disease
EHR	electronic health record
EHIF	Estonian Health Insurance Fund
FD	family doctor
FDA	US Drug and Food Administration
HGP	Human Genome Project
HIS	hospital information system
IM	Individualized Medicine
MO	medical oncologists
NHS	National Health Services
PM	personalised medicine
POG	Personalised Onco-Genomics
PGT	personal genomic testing
SNPs	single nucleotide polymorphisms
WGT	willingness to pay
WHO	World Health Organization
WGS	Whole-Genome Sequencing

Table of Contents

List of figures

List of tables

1 Introduction

Cardiovascular diseases are the main cause of death in Estonia and worldwide [1]. According to the latest Eurostat Statistics, the total number of CVD deaths for EU was 126,3 per 100 000 inhabitants and in Estonia there were 295,5 deaths per 100 000 Inhabitants and for every 100,000 residents, at least 250 men and 80 women under the age of 65 die of CVD each year. There are also found gender-based differences in developing CVD when looking at genetics and its possibilities and therefore they should be handled differently [2]. CVDs have a negative socio-economic impact because mostly, people in the working age develop CVD. The Estonian numbers of CVD incidents are significantly higher than in other developed countries and could be reduced by (more) efficient preventive work [3]. The study done by Kaldamäe et al in Estonia also proved that there is a high prevalence of cardiovascular disease risk factors in Estonian adults (20-65 years of age) [4].

According to the European Guidelines on cardiovascular diseases prevention, CVD remains a leading cause of morbidity and mortality, despite the improvements and in outcomes. The age-adjusted coronary artery disease (CAD) mortality has declined over the years, especially in the high-income countries [5]. Moreover, CVD represents a considerable economic burden to society and effective preventive measures are necessary. For example in 2009, the costs related to CVD totalled €106 billion in the European Union, making ~9% of the total healthcare expenditure across the EU [6].

There is consensus in favour of an approach combining strategies to improve CV health across the population at large from childhood onward, with specific actions to improve CV health in individuals at increased risk of CVD or with established CVD.

Cardiovascular disease (CVD) was also the leading cause of death in the United States in 2016, accounting for more than 900 000 deaths [7]. Despite large declines in CVD mortality in the late 20th century attributed to advances in public health and health care,

improvements in US life expectancy have slowed for some groups, and CVD mortality is no longer improving.

Studies show that the cardiovascular diseases are highly preventable if detected early and started with right treatment early. Cardiovascular diseases are often considered as lifestyle disease and with appropriate nutrition and lifestyle change often preventable and in of case already started disease the life conditions can be improved [8].

Since recent years, and specifically in 2003, when in the US the Human Genome Project (HGP) was successfully completed and since the human genome sequencing costs are going down, the new medical approach called Personal Medicine has emerged. After that, the Personalised Medicine trends have been followed also in Germany, UK, France, Norway and in other countries. It has also been found according to Jain, Kewal, who researched about the personalised management of cardiovascular disease, that several cardiovascular diseases are recognized to have a genetic component, which means that through genetic testing the genes associated with cardiovascular diseases could be defined and listed [9].

According to the current urgency of the topic and the number of people and their families affected in Estonia, it is important to study the views of population as well as health professionals' views about the acceptance of genetic testing.

The aim of the research is as follows:

- To assess the acceptance and understanding of the genetic information by population and health professionals in Estonia in the CVD prevention process.

Current theses is composed of two main parts. First part gives a theoretical background of the research subject. Second part demonstrates the research conducted, its objectives and questions, methodology, results of the study, discussion and summary.

2 Background and definition of cardiovascular diseases

In this chapter, the author clarifies the definitions and descriptions of what are cardiovascular diseases (CVD), what are risk factors of cardiovascular diseases, what is genetic risk score of CVD, what is personalised medicine and what research has been done until now in the field of population and health professionals' acceptance and views about genetic testing.

2.1 What are cardiovascular diseases?

According to the World Health Organization definition about cardiovascular diseases, the cardiovascular diseases (CVDs) are a group of disorders of the heart and blood vessels [1]. Depending on the place of the disease in the blood vessels, CVD includes following diseases:

- coronary heart disease (a disease of the blood vessels supplying the heart muscle);
- cerebrovascular disease (a disease of the blood vessels supplying the brain);
- peripheral arterial disease (a disease of blood vessels supplying the arms and legs);
- rheumatic heart disease (damage to the heart muscle and heart valves from rheumatic fever, caused by streptococcal bacteria);
- congenital heart disease (malformations of heart structure existing at birth);
- deep vein thrombosis and pulmonary embolism (blood clots in the leg veins, which can dislodge and move to the heart and lungs).

Acute events like heart attacks and strokes are associated with changes in the blood vessels (blockage that prevents blood from flowing to the heart and brain). The cause of attacks and strokes is usually a combination of several risk factors such as unhealthy diet, sedentary lifestyle, and excessive use of alcohol, hypertension, diabetes and hyperlipidaemia (too high concentration of triglycerides or cholesterol in the blood).

2.2 What are risk factors for cardiovascular diseases

As stated in WHO information sheet about CVD, research study done by Jack, Stewart et al the common behavioural risk factors of heart disease and stroke are physical inactivity, unhealthy diet, tobacco use and harmful use of alcohol. Because of the abovementioned behaviour, some individuals will have raised blood pressure (BP), raised blood glucose, raised blood lipids, overweight and obesity [1], [10]. In addition to the behavioural risk factors, according to WHO, 17.7 million people die each year from CVD, which is an estimated 31% of all deaths worldwide. More than 75% of all CVD occur in low-income and middle-income countries [1]. Newer studies done on CVD and their causes bring out that next to behavioural risks there are for example family history (FH) to take into consideration as well as personal genes. For example, literature research done by Imes and Lewis shows, that FH can contribute to the persons CVD related risk factors [11].

However, further research needs to be conducted since a person's awareness of their FH of CVD or their own risk for CVD is not a sufficient predictor of changes in their health-related behaviour. If we look at studies done about personal gene influence or genetics, then for example according to Kathieresan and Srivastra in the general population, a history of premature atherosclerotic CVD in a parent can increase the CVD risk in their offspring about 3.0-fold. It is also to note, however, that the exact magnitude of the role of inheritance, varies by disease and by other factors such as persons age when the disease started and subtype of disease. Healthy aging is something that has been researched and how to understand the most dominant factors of developing CVD [12]. Therefore, family history of early-onset of CVD has long been considered as a risk factor for the disease and does contribute to increase the risk independently and in addition to the well-known risk factors [13].

Nadar, et al conclude in their research, that there is not one gene on its own that leads to CVD, but it is rather an interaction between the effects of various genes. Single nucleotide polymorphisms (SNPs) are single base changes in an individual's genome that differ from the usual base at that location. To have more substantial and more precise results, a large number of candidates SNPs should be tested in a longitudinal long-term study. It can be concluded, that currently, there is still a long way to go to apply the genetic knowledge to either prevention or treatment of CVD. However, by more research and understanding

of these genes and the genetic basis of various risk factors would still enable us to identify high-risk populations, to undertake primary prevention or prophylactic measures [14].

Several recent genome-wide association studies have identified candidate genes associated with CVD. Since the effect of each genetic polymorphism is small, most studies have used genetic scores to summarize the genetic component. There is currently lack of consensus regarding which genes and their corresponding single nucleotide polymorphisms (SNPs) should be included in a genetic risk score and which method should be used to calculate the genetic score [6].

Currently, many commercial tests are available; allowing an almost complete assessment of an individual's genome, and strong pressure is being applied to use this information to predict genetic risk and to make genetic testing a routine measure. Given the lack of agreement regarding which genetic markers should be included, how genetic risk scores should be calculated and uncertainties about improvement in CV risk prediction, but interestingly enough, the use of genetic markers for the prediction of CVD is not recommended according to Singleton et al [15].

2.3 Genetic risk score and CVD

A polygenic score, also called as polygenic risk score, genetic risk score or genome-wide score is number based on variation in multiple genetic loci and their associated weights. It serves as the best prediction for that trait that can be made when considering variation in multiple genetic variants. The benefit of the polygenic score is that according to them, future can be predicted, for example future diseases susceptibility like CVD. According to Paquette et al, although familial hypercholesterolemia (FH) is a severe monogenic disease, it has been shown that clinical risk factors and common genetic variants can modify cardiovascular disease (CVD) risk. They conclude in their recent research that even in the context of a severe monogenic disease such as FH, common genetic variants can significantly modify the disease phenotype.

The use of the specific SNPs (192-SNPs GRS_{CAD}) may refine CVD risk prediction in FH patients and this could lead to a more personalised approach to therapy [16]. See also Figure 1 below.

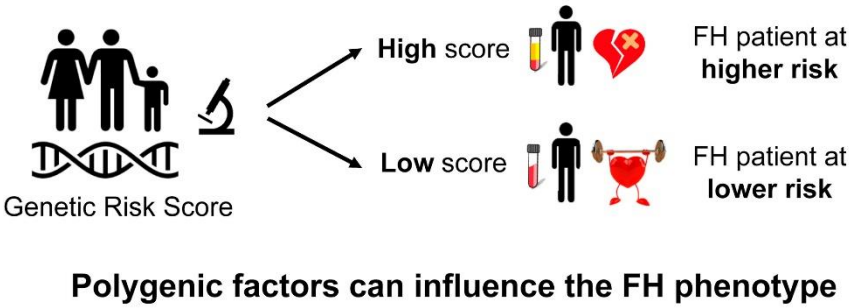

Figure 1. Polygenic factors can influence the FH phenotype

Hayato et al researched atrial fibrillation (AF) and how to identify the people at risk throughout their lives. They investigated, whether multiple single nucleotide polymorphisms together as an AF genetic risk score (AF-GRS) can improve prediction of one's risk for AF and as a result they concluded that an AF-GRS can identify 20% of individuals who are at 2-fold increased risk for incident AF and at 23% increased risk for ischemic stroke. They concluded that targeting diagnostic or therapeutic interventions to this subset may prove clinically useful [17].

According to Jain, Kewal, who researched about the personalised management of cardiovascular disease, concluded, that several cardiovascular diseases are recognized to have a genetic component, therefore the CVD prevention can be addressed from personalised medicine point of view [18].

All in all, it can be concluded that the use of risk factors for decision-making in cardiovascular disease has already a long history in medicine. At the beginning, when first attempts to use genetic risk scores in addition to the traditional risk factors failed due to too little understanding of the genetic basis of the complex cardiovascular disease then today, the topic is more understood and researched. However, newer studies show, that genetic risk scores can now outperform traditional risk factors in risk prediction. The time is ready to start incorporating genetic risk scores into clinical practise [19].

However, according to Schee et al: “Personalised medicine (PM) is no longer an abstract healthcare approach. It has become a reality over the last years and is already successfully applied in the various medical fields. Although there are success stories of implementing PM, there are still many more opportunities to further implement and make full use of the potential of PM [20]. And the question would remain, whether the precision medicine would be the route to a healthy world [21]?

2.4 Recent studies about population acceptance of genetic testing

Technologies have changed; new methods and possibilities have emerged in medicines different aspects and branches. The topic of genetic testing is closer and more accessible to the population as is ever has been before. In many countries all over the world, the issues of genetics are on the table. Would we have success in that? Would the population or health professionals accept the new challenges associated with the novelty and accessibility of the testing?

There is some recent research about community engagement in genetics, and to cite the research of Etchegary et al: “With the proliferation of biobanks and the rapid pace of discoveries in genomics research, public support will be crucial to realize health improvements. If researchers can engage the public in regular, transparent dialogue, this two-way communication could allow greater understanding of the research process and the design of efficient and effective genetic health services, informed by the public that will use them” [22].

2.4.1 Current research on population views and genetic testing

Recently, there was a study done by Ostergren et al about “How Well Do Customers of Direct to Consumer Personal Testing Services Comprehend genetic test Results?” The research was done on the studying the customers of two companies 23andMe and Pathway. The aim of the study was to assess customer comprehension of health-related personal genomic testing (PGT) results. In conclusion, most customers did interpret accurately the health implications of PGT results; however, comprehension varied by demographic characteristics, numeracy and genetic knowledge, and types and format of the genetic information presented. Results suggest for a need to tailor the presentation of PGT results by test type and customer characteristics [23].

Marshall, D, et al concludes in her study, that yes, the prices are going down for whole-genome sequencing, but who will have the access to it and who can afford it? The study results show, that there is more interest from the people who are younger, highly educated and have higher paying jobs. Sex, age, education, income, genomic knowledge and knowing someone who had genetic testing or having had genetic testing done personally were associated with significantly higher whole-genome sequencing (WTP) for willingness to pay (WGS). After controlling for income and education, males were willing to pay more for WGS than females [24].

Etchegary et al came to a conclusion in the study done in members of the general public in Newfoundland Canada, that 87.01% participants of the survey strongly agreed that if genetic test showed that they were at increased risk for a disease, it would affect the decisions they would make about their health [22].

Lee at al did research on public awareness in Korea and performed a self-administered questionnaire survey on member of the public, who participated in the survey on a voluntary basis. The idea was to measure public knowledge, attitude, and acceptance of PM. They found, that only 28% of participants had knowledge that genetic factors can contribute to inter-individual variations in drug response and the definition of PM. Higher family income was correlated with greater knowledge about PM. Most respondents preferred integrated pharmacogenomic testing over drug-specific testing and agreed to inclusion of pharmacogenomic testing in the national health examination, but only 51% were willing to pay for it. Their results identify the urgent need for population education as well as the potential health disparities in access to PM [25].

Gastrow et al investigated recently the population views of South Africans about the various aspects of biotechnology, including genetics, genetic testing, gene therapies. The results showed, that younger generations were more positive about a variety of biotechnology-related issues and the overall risk/benefit assessment of biotechnology. Understanding the causes of those correlations presents an objective for future research. For example, the concepts of DNA and genes are reportedly far better understood than those of genetic modification or GM food and would therefore present a better starting point for engagement and knowledge transfer. Higher education and living standard was associated with greater knowledge about the topic and their personal positions about it. It

is not note, that when it was asked about genetic testing to detect inherited diseases, then about 49% of the respondents had not heard of it, and 28% had heard something about it but had very little information about it. To the question about gene therapies to treat genetic conditions – 52% had not heard of it and 25% had very limited knowledge about it what it really was. One of the conclusion of the studies was that whatever the causes, the implication is that the future South African public is likely to be more knowledgeable about biotechnology and have more sharply defined attitudes towards biotechnology [26].

2.4.2 Physicians attitude and knowledge about genome data usage

Chow-White, P, et al investigated about the medical oncologists' (MOs) genomic literacy and their experiences based on their participation in a cancer genomics trial in British Columbia, Canada. The results show a low to moderate level of genomic literacy among MOs. There were geographical differences in the results: MOs located outside the Vancouver area (the major urban centre) reported less knowledge about new genetics technologies compared to those located in the major metropolitan area (26.7 vs 73.3%, $P < 0.07$, Fisher exact test). Forty-two percent of all MOs thought medical training programs do not offer enough genomic training. Most of the respondents thought genomics will have major impact on drug discovery (67.7%), and treatment selection (58%) in the next 5 years. They also thought the major challenges are cost (61.3%), patient genomic literacy (48.3%), and clinical utility of genomics (42%). The data suggest that there is a high need to increase genomic literacy among MOs and other doctors in medical school training programs and beyond, especially to physicians in regional areas who may need more educational interventions. Initiatives like Personalised Onco-Genomics (POG) play a critical role in the education of MOs and the integration of big data clinical genomics into cancer care [27].

2.5 About personalised medicine

The usage of the term personalised medicine came to usage in 2003 when first genomic tests took place in the framework of Human Genome Project (HGP) and it has been started to talk about personalised approach in patients more suitable treatment selection. The main idea behind genome sequencing is to find out the most appropriate treatment for a patient in case needed. In comparison – wrong medication or medications not accepted

by a person's body may lead in increase in hospitalizations, higher payment for health insurance funds (when wrong diagnosis is given, and new drugs need to be prescribed and tested) and decrease in death (having to know the perfect amount of medication needed to treat one person). The term personalised medicine has many names and terms in different countries. It is called precision medicine, personal medicine, individualized medicine, stratified medicine. The tailoring of treatment to patients dates to the time of Hippocrates, but the terms has come to usage in the resent years with the growth of new diagnostic and informatics approaches, particularly due to genomics [28].

2.5.1 Personalised Medicine in Germany, UK and U.S.

The U.S. and Germany are brought as examples here, since they are frontrunners in the personalised medicine. Kichko et al did a survey comparing the state of personalized medicine in Germany and U.S. The successful completion of the Human Genome Project (HGP) in 2003 and the fast decreasing human genome sequencing costs encouraged the development of a new medical approach which is called Personalised Medicine. The term personalised Medicine (PM) has been in use for about a decade and depending on the scope, its definition varies widely. The Personalised Medicine Coalition defines Personalised Medicine as "the use of new methods of molecular analysis to better manage a patient's disease or predisposition to disease''. The FDA defines Personalised Medicine (also called as "precision medicine") as an innovative approach to tailoring disease prevention and treatment that takes into account differences in people's genes, environments, and lifestyles [29].

Personalised Medicine has gained more attention not only in the U.S. but also in Germany, where it is better known as Individualized Medicine (IM). By the end of 2010 the German Federal Ministry of Education and Research (BMBF) named Personalised Medicine to be one of the six priorities and introduced the Action Plan called "Individualized Medicine: a New Way in Research and Healthcare" [29].

As the largest country-specific PM projects in Europe outside Germany, the projects in the UK, France and Norway can be named. In the UK, in 2011, the Stratified Medicine Initiative was launched, with a £60 million budget, which focused on patient cohorts and biomarker, genotypic and phenotypic analyses. In 2012 Genomics England launched the

100,000 Genomes Project. The goal of the project was to collect genome sequencing of 100,000 patients by 2017. In France there were the Integrated Cancer Research Centres projects (SIRIC) and the French National Alliance for Life Sciences and Health projects (Aviesan). The SIRIC projects were designated to find new opportunities for cancer treatment. In Norway, a so called HUNT study was to combine genetic data with clinical records, cancer, stroke and death registries [29].

While designing the questionnaires to their studies, Kichko et al pre-defined some hypotheses which they wanted to prove. They assumed that Personalised Medicine is enhanced by the problematic of adverse drug side effects. They also predicted that healthcare participants in the U.S. would desire more patients' involvement in the decisions about their medical treatment, as is in Germany. As Personalised Medicine comes from the U.S., they believed that the concept as a whole, as well as personalised drugs and pharmacogenetic tests, would be better known, accepted and used among the public and physicians in the U.S. rather than in Germany. Additionally, they assumed that the public PM acceptance depends on age, gender and health insurance availability and its coverage. They also expected that the physicians working at the hospitals would be better informed about the advantages of Personalised Medicine, and it was suggested that less than 10% of physicians had sufficient experience in analysing the results of genetic and pharmacogenetic tests today, and that physicians' willingness to get trained on PM depends on their age. Physicians having Electronic Health Records and family medical histories of their patients were seen to be more likely to accept Personalised Medicine [29].

In conclusion, all in all, the public and physician opinions on Personalized Medicine aspects and preconditions for the wide implementation in Pennsylvania and Bavaria were not as different as they originally expected them to be. Most of the respondents had concerns about adverse drug side effects and wished to increase patient involvement and standardize medical regulations. There was a common opinion about the genetic data exchange, date it was not secure and, that the offer of personalised drugs and tests online should not be increased and there should not be one centralised genetic database managed by the government [29].

According to Jain, Kewal, who researched about the personalised management of cardiovascular disease, several cardiovascular diseases are recognized to have a genetic component. Both genetic and lifestyle factors are key drivers of coronary heart disease (CHD). Genome-wide association analyses have identified >50 independent loci associated with the risk of CHD. Quantification of both genetic and lifestyle risks in 3 prospective cohorts and 1 cross-sectional study revealed that adherence to a healthy lifestyle was associated with a substantially reduced risk of CHD within each category of genetic risk. A healthy diet, exercise, smoking cessation, and stress reduction are recognized measures for reducing the risk of cardiovascular disease. A study compared the effectiveness of these lifestyle interventions for individual risk factors for reducing the 10-year cardiovascular disease risk and found yoga and smoking cessation to be the most effective forms of cardiovascular disease prevention. The benefit is enhanced when interventions are performed simultaneously rather than as single measures [18].

2.5.2 Personalised medicine in Estonia

The Estonian Biobank was founded in 2000 as a population-based biobank. A decade later, the biobank includes a collection of health and genetics data of around 5% of the adult population of Estonia. Due to the Human Genes Research Act it is allowed to regular updating of data through linkage to national registries enabling long-term follow-up of the cohort. In addition to promoting the development of genetic research, the Estonian Biobank data has used data available in the Estonian Biobank for a wide variety of research projects nationally and through international collaborations. In the past few years increasing amount of attention has been placed on translating the results of genetic research to improve public health. In 2014, the Estonian Government supported a plan for a shift toward precision medicine based on modern genetic technology [30].

In 2018, Estonia has started a program to recruit and genotype 100,000 new biobank participants as part of its National Personalised Medicine programme. The Estonian government plans to develop its healthcare system by offering all its residents genome-wide genotyping that will be translated into personalised reports for use in everyday medical practice through the national e-health portal. The country has currently many encrypted digital solutions incorporated into government functions that link the nation’s various databases through end-to-end encrypted pathways. The initiative is a joint

development project of the Ministry of Social Affairs, the National Institute for Health Development and the Estonian Genome Centre of the University of Tartu [31].

As stated in earlier literature research and other international efforts have identified thousands of associations between genetic variants and diseases or traits and created maps of the unique variation within populations [32], [33], [34].

3 Research objectives and questions

The main aim of this study is to demonstrate the willingness to use genetic testing results and the expectations of the population and the health professionals as a CVD preventive measure in the personalised medicine approach.
The study is aimed at demonstrating the acceptance and understanding of members of the population and health professionals' attitude towards personalised medicine from the point of view on CVD prevention.

The aims include:

- To assess the acceptance and understanding of the genetic information by the population and health professionals.

The author has set the objectives of this thesis project as shortly described below:

- To study the willingness and interest of public and health professionals to have the prevention activities of cardiovascular diseases include genetic testing data and personalised medicine approach.
- To investigate the formats and channels through which the population and health professionals would like to receive access to genetic testing results.
- To propose 3 – 4 changes in the cardiovascular disease prevention strategy from the personalised medicine point of view.

Accordingly, the author has formulated the following research questions.

- What is the acceptance and understanding of the genetic information by the population and health professionals?
- In what form, from whom or through which channels would the population and health professionals be expecting to receive access to genetic testing results or health data?
- What is the role of health analytics, artificial intelligence (AI), health coach and patient summary in CVD prevention process?
- What would be the 3 – 4 possible changes proposed to the primary care intervention for CVD prevention from the personalised medicine point of view?

Research hypothesis:

- Population is ready to have genetic data included into the prevention process of CVD and are expecting primary care level consultations according the personalised medicine point of view.
- Health professionals are willing to use the benefits of personalised medicine and accepting genetic data for broader patient health information availability.
- Population and health professionals are ready to accept novel information receiving formats - health summary by the health coach or health information analysed by AI or health analytics.

4 Methods

In the current study, there will be two online surveys conducted to gain insight into the current views of the population and health professionals. The surveys will be conducted to understand the participants' views about personalised medicine approach in CVD prevention. Descriptive quantitative research methods have been used.

After the analysis and description of the current trends and views, 3 – 4 possible changes in the cardiovascular disease prevention process according to the personalised medicine and the inclusion of patients' genetic data will be proposed.

4.1 Research methodology

Two surveys will be conducted, one to get the opinion of the population side and in the other health professionals opinion about personalised medicine, gene testing and the possibility of patient summary and analytics will be asked.

The population survey will include questions about person's background (age, sex, educational background), about their knowledge about CVD and family history of CVD, their opinion about genetic testing and health analytics and AI, and how they would like to receive information about their health state. For the question formation, a Stanford questionnaire will be used and modified [35].

The questionnaire towards population will have 10 questions grouped into six sections:

- General background: education, age, sex
- Opinion and knowledge about family history and own CVD
- Opinion about their own health condition
- Question about regular physical activities
- Opinion about how and in what form to get information about current health state
- Attitude towards genetics and prevention behaviour.

The health professionals' survey will include 10 questions, including questions about their organisations willingness to investigate issues of personalised medicine, gene testing and the doctor's personal willingness and interest in consulting their patients form the point of view of personalised medicine.

4.1.1 The expected outcomes of the population surveys results

Predictable or expected result of the population survey are following. Population is willing to have genetic data integrated into their health account and they would more likely be willing to make lifestyle changes in case when genetics is involved, and their personal genetic data is known to them. However, genetic data needs to be analysed and interpreted for them and presented in a suitable and understandable way for them. Health literacy skills is something we should consider when designing preventive materials and information for the people. Studies show that there are above 90 Million people in the United States, whose health literacy skills are not adequate [36]. Health literacy skills are something that has been researched extensively and Cusack et al concluded that educational interventions are necessary, to improve people's understanding of key concepts for evaluating health intervention claims and can improve people's knowledge and skills, if not for a longer period of time, but at least in the short term [37]. It has also been researched that higher health literacy scores were associated with less CVD risk such as lower body mass index (BMI), less metabolic syndrome in women, and less fatty liver disease [38]. It has also been researched that by educating the population over a period of time, then treatment compliance and understanding of the need for treatment would improve [39].

According to current study results in this thesis, population is generally interested in receiving information about their health state from a health coach (instead of the primary care physician) and would not mind receiving an analysed summary of their health state.

4.1.2 The expected outcomes of the health professionals' surveys results

The author expects from the health professionals survey results that the health professionals would be more sceptical towards the usage of genetic data than the population, but they would be willing to learn about genetics and welcome it in their daily working routine. They would be interested in receiving a patient summary from a health coach or data analysed by AI, rather than analysing the data by themselves. Health analytics is a positive topic for them and they would welcome it, if their everyday work would become easier due to that.

4.2 Research sample

For the population study group views and opinion, the people aged 18 – 50 were selected from the whole group of survey respondents. The reason for excluding older participants was that the aim of the study was to analyse this age group as it was assumed that they are more willing to accept genetic testing and they are in the age group, where prevention (rather than treatment) is still possible.

For the health professionals' views and acceptance, all respondents' views were collected and analysed and separately, the cardiologists and general practitioner views were analysed and brought out separately since they were first point of contact for people being involved with CVD prevention process.

Population survey was shared among internet users aged 18+ via Facebook and through personal contacts via electronic channels. Survey was also allowed to be shared among participants on their Facebook pages or privately. About 100 patients were to be expected to answer the questions. The total number of respondents was 81 and in the selected age group for this study, the number of respondents was 63 people. The survey was distributed via electronic channels due to the fact, that younger, 18 – 50 years of age peoples' opinions and views were expected. The reason for selecting younger internet users' responses for this study was, since literature research has shown that for the example of the study of the companies 23andMe and Pathway was stated, younger people are more likely to accept genetic testing [23].

Health professionals' survey was shared via e-mail in East Tallinn Central Hospital, North Estonia Medical Centre Foundation, and some selected general practitioners' offices in Tallinn such as "Perearstikeskus Sinu Arst", "Tallinna Munitsipaal Perearstikeskus", "Meditiim Perearstikeskus", "Järveotsa Perearstikeskus", "Pirita Perearstikeskus". About minimum 10-15 survey answers were expected to be collected, but the total number of collected responses was 31.

4.3 Data collection

Data was collected in two separate study groups and therefore it will be described separately below.

4.3.1 Population opinions collection

For the data collection from the written surveys, general data was collected about age, sex, education level. In addition, information was collected, whether they were aware of their family history and own existing CVD, their opinion about their own health condition, question about the amount of weekly regular physical training, opinion about how and in what form to get information about their current health state and their attitude towards genetics and prevention behaviour.

4.3.2 Health professionals' opinions collection

For the data collected from the written surveys, following information was collected: information about their profession, their opinion about personalised medicine and their institutions view about it and whether they know that it is planned to integrate the genetic data into their hospital information system. It was also asked whether they would see value in CVD preventive activities in having access to patients' genetic data and whether they would have the knowledge how to interpret the patients' genetic information. In addition, data was collected about the way health professionals would like to have access to the patients' data and whether the health professionals would see any benefit of patients own data collected via activity monitors and such.

4.3.3 Data gathering

Data gathering, which was divided into two phases (described in detail under 4.3 Data Collection):

- data obtaining from the population surveys,
- data gathering from the health professionals' surveys will be collected.

4.3.4 Data analysing

The population data was analysed after the collection and grouped into several categories such as the answers by age, by education, by sex. In total results, the responses were grouped into one category – age group of 21 – 50 years old participants.
The health professionals' data was analysed after the collection, and the opinions were grouped by profession: all answers, answers by cardiologists and answers by family physicians.

5 Results

The study was conducted, and the data was collected electronically in the timeframe 04.04.2018 – 30.04.2018 from the respondents' survey results. Two surveys were conducted – one addressed towards general population and the other towards health professionals (cardiologists, general practitioner and other medical specialists or other medical personnel working in the hospital or family physician's centres). In the population surveys, from the total respondents aged 18 – 80, subjects aged 21 – 50 years were selected. Data was collected electronically from the survey responses and analysed. There were no respondents in the age group 18 – 20 years old and subjects 51 – 80 years old were excluded, since they did not meet the selection criteria. There were no respondents in the age group 81+ years. The total number of people excluded was 18 subjects (22%) from the total respondents. In the population view group, totally the data of 63 survey respondents was selected for analysis.
In the health professionals group, all respondents' answers were collected and analysed. The total number of 31 subjects was included and analysed for this research.

Results were divided into two subgroups:

- Results – population
- Results – health professionals

5.1 Results – population

From the total number of 81 subjects 63 subjects were in the selected group. The division between the sexes was following: 8 subjects (12.70%) were male and 55 subjects (87.30%) were female. There were 16 people (25.40%) in the age group 21 – 30 years, 20 people (31.75%) were 31 – 40 years old, and 27 people (42.86%) were in the age group 41 – 50 years old. Educational background of subjects was 53 people (84.13%) had bachelors or master's degree, 4 people (6.35%) had secondary education and 6 people (9.52%) had a postgraduate degree.

Table 1. Profile of selected subjects for population view

Variables	Number of participants	Percentage (%)
Male	8	12.7
Female	55	87.30
21 – 30 years	16	25.40
31 – 40 years	20	31.75
41 – 50 years	27	42.86
Secondary education	4	6.35
Bachelors or master's degree	53	84.13
Postgraduate degree	6	9.52

In the study group, to the question, how would they rate their current health state, 4 people (6.35%) rated their current health state as "excellent", 30 people (47.62%) as "very good", 21 people (33.33%) as "good", 7 people (11.11%) as "fair" and 1 person (1.59%) as "poor". Totally, from 63 subjects, 8 people (12.7%) stated their health being "fair" or "poor" and other 55 people (87.3%) as "good", "very good" or "excellent".

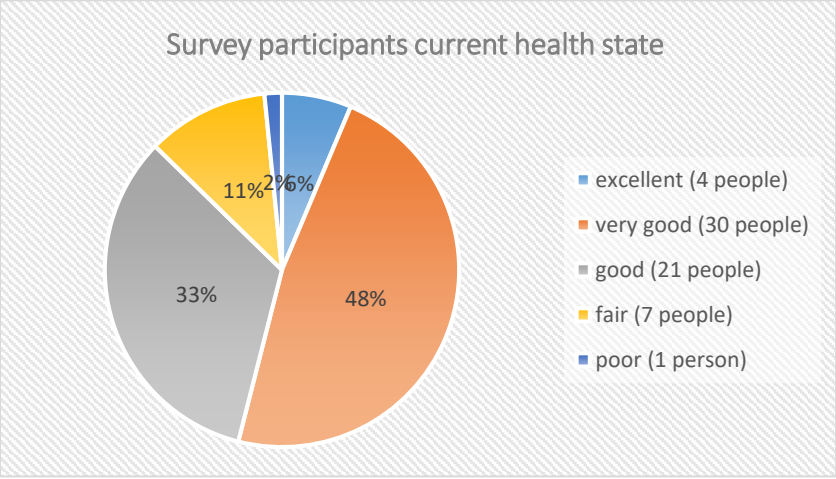

Figure 2. Survey participants own health state perception

To the question whether the subjects did have any cardiovascular disease (high blood pressure, obesity or hypertension), 47 people (74.60%) stated that they do not have and CVD and 3 people (4.76%) did not know, if they had any cardiovascular diseases. 13 people (20.63%) however were aware of their own current cardiovascular diseases.

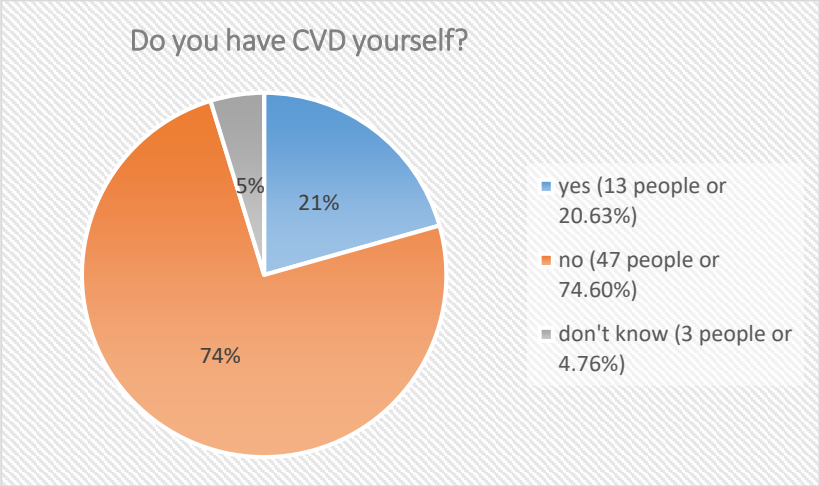

Figure 3. Participants own current cardiovascular diseases

Since in genetics, often family history plays a role, it was asked, whether the subjects knew about their family history about Diabetes 1 or 2. 41 people (65.08%) stated that their parents or grandparents did not have any such diseases, 18 people (28.57%) mentioned that "yes", there was a family history (FH) in diabetes 1 and 2 and 4 people (6.35%) answered that they did not know if such diseases existed in their family.

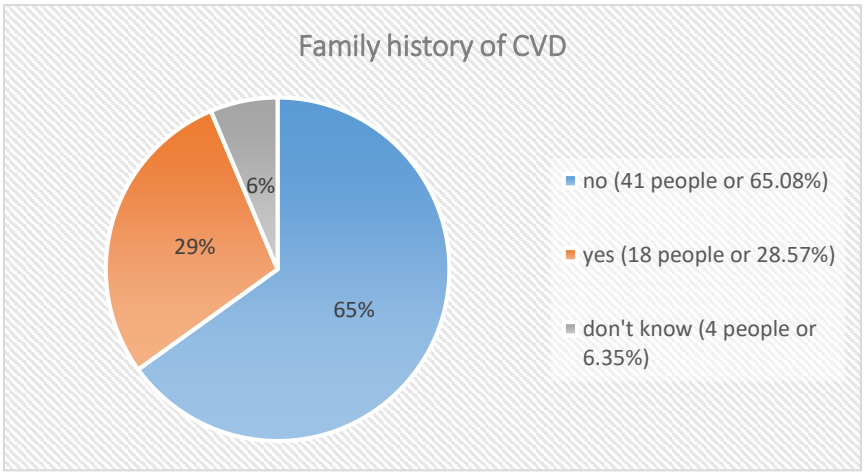

Figure 4. Family history (in parents and grandparents) of CVD

As one of the preventive activities towards a healthy heart according to WHO recommendations [1], and other relevant guidelines, moderate to active regular (weekly based) physical activity is suggested. Therefore, it was asked from the survey participants, how much of any physical activities a week they would do. The activities listed in the survey question were brisk walking, jogging, bicycling, and other relevant activities. For the selection of weekly amount of activities following options were given: 0 minutes a

week, less than 30 minutes a week, 30 – 60 minutes a week, 1 – 3 hours a week, 3+ hours a week. The majority, 24 people (38.10%) of the respondents replied, that they did sports at least 1-3 hours a week, 17 people (26.98%) did sports for 3+ hours a week, 16 people (25.40%) did sports for 30 – 60 minutes a week. 1 person (1.59%) stated that he/she did not do any sports a week (0 minutes a week) and 5 people (7.94%) did sports less than 30 minutes a week. In total, 41 people (65.08%) did more than 1 hour a week sports and 22 people (34.92%) less than 1 hour a week.

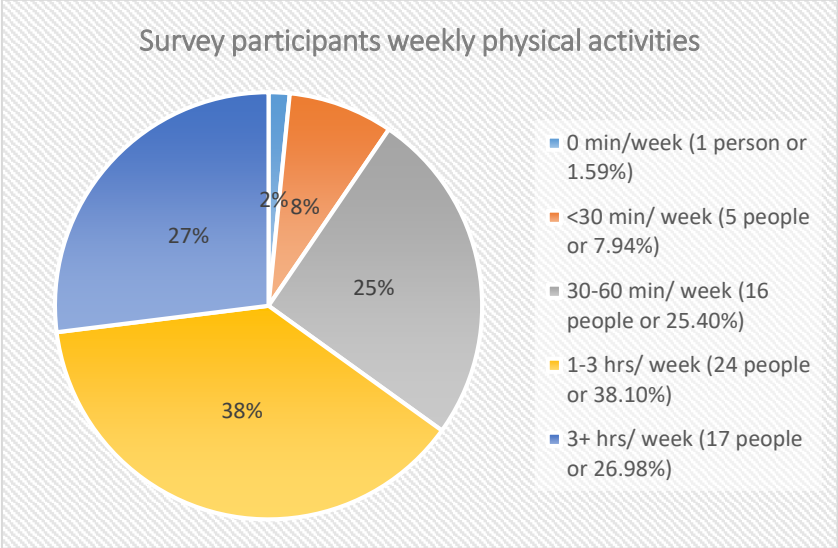

Figure 5. Survey participants' weekly physical activities

As one of the study objectives was to investigate what would be the formats and channels through which the subjects (population) would like to receive access to genetic testing results, it was asked about the preferred personal health data accessing formats. In addition, the idea of having regular access and update of current health stat to keep track of own health state and plan whether the preventive or treatment activities are needed, it is important to know the current state of personal health. Therefore, it was asked about, how people would like to get an opinion about their current health state.

For the access of the information in patient portal, 5 people (7.94%) choose the answer, to have the information available from My Health account, 6 people (9.52%) chose it as the best option for them. 16 people (25.40%) stated that receiving information from their family doctor is a sufficient choice for them. 12 people (19.05%) stated that it would be good, if artificial intelligence would analyse their health data and the summary of it would be made available to them in their health account. 21 people (33.33%) answered that they

would like to receive the information from the health coach, who would analyse their data and give feedback to them. 3 people (4.76%) replied that they can handle their health state by themselves. No one chose the option "other", where the respondents could specify their answers.

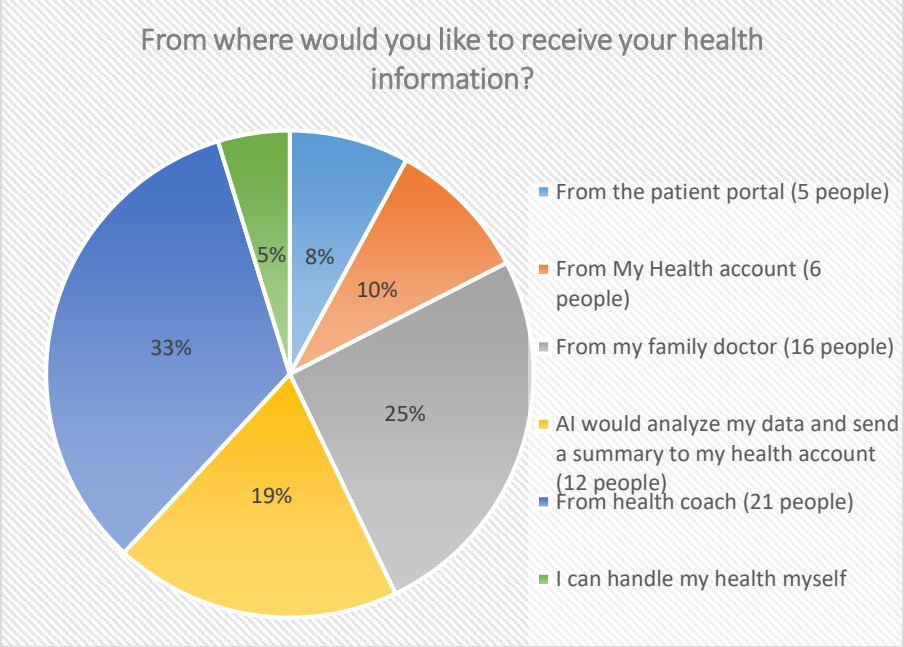

Figure 6. Populations preferable format of health status access

If we summarize the electronic sources (information received from electronic formats such as patient portal, my health account and AI) vs personal sources (feedback from family doctor of GP), then we can see that most of survey respondents (70%) would favour receiving information in electronic format and in summarized way.

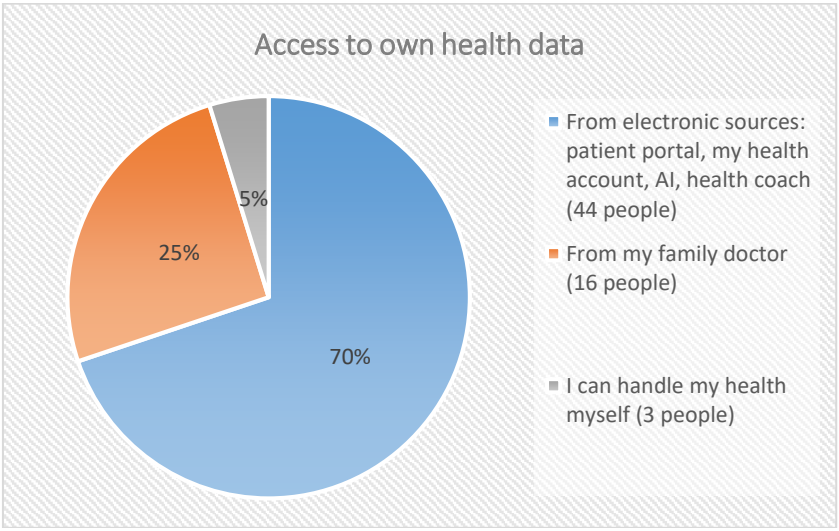

Figure 7. Populations preferable access to health data

In the last two questions it was asked, whether the respondents would trust their family doctor more than genetics. The questions were following: “If your family doctor would tell you that you are overweight, and you would benefit from losing weight, would you do so?” 42 people (66.67%) replied that “yes, I would do it”, 20 people (31.75%) replied that “yes, I would consider it” and 1 person (1.59%) replied that “I know it is important, but don’t see a benefit in that”. No one replied, “Not sure it is important” or “definitely no”.

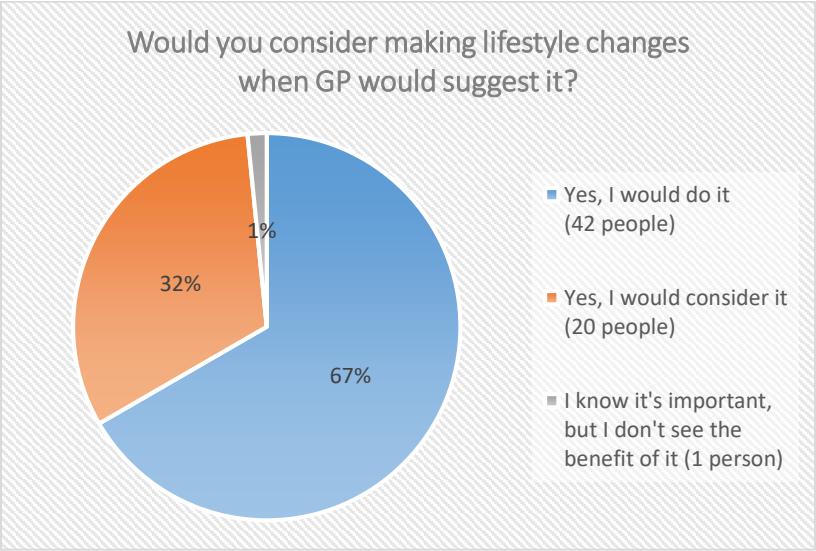

Figure 8. Populations lifestyle changes when GP would suggest

Last question was addressed towards changing lifestyle when genetic testing results were involved and was stated as following: “Studies show that 30% of our health is influenced by our genes. If you would get to know (through gene testing) that you belong to the high-risk behaviour class in developing any heart diseases, would you then consider changing your lifestyle to prevent the onset of a disease or to gain few life years?”
44 people (69.84%) replied, that “yes, definitely”, 16 people (25.40%) did reply “maybe”, 1 people (1.59%) replied “no” and 2 people (3.17%) replied that “I don’t believe in genetics”.

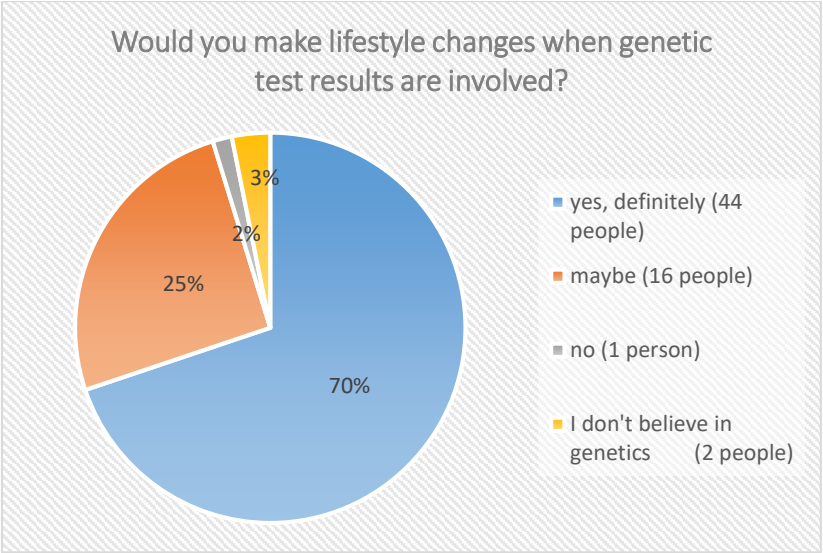

Figure 9. Possible lifestyle changes when genetics is involved

5.2 Results – health professionals

In total, there were 31 answers collected and the specialities of the participating health professionals were following: 12 cardiologists (38.71%), 5 other medical doctors (16.13%), 4 family doctors (12.90%), 4 nurses (12.90%) and 6 other medical personnel (19.35%).

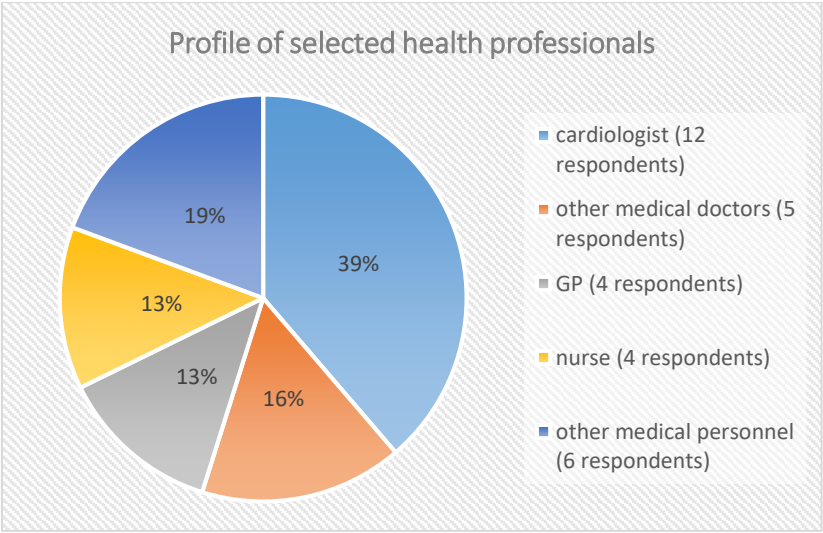

Figure 10. Profile of selected health professionals

To the question “What do you think about the personalised medicine?”, total of 22 respondents (70.97%) replied that “it is important”, 1 person (3.23%) replied that “it is not important”, 5 people (16.13%) replied that ”it is very much needed” and 3 people (9.68%) replied that “it is somewhat needed”. Totally, only 1 person (3.23%) mentioned that “it is not important” and the rest of the respondents, 30 people (96.77%) said that

personalised medicine is “somewhat important”, “important” or “very important”. None of the respondents mentioned that the personalised medicine is not important.

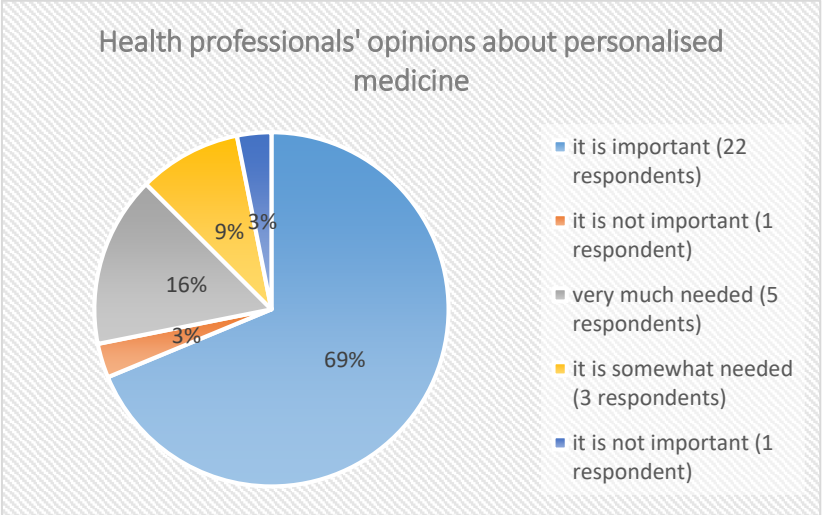

Figure 11. Health professionals’ thoughts about personalised medicine

On the question whether personalised medicine would play an important role in the next 5 years in your organisation, 13 people (41.94%) replied that yes, “it will play a significant role” and 13 people (41.94%) that “it will play a moderate role” and 5 people (16.13%) replied that “the role of personalised medicine will be small” in their institutions.

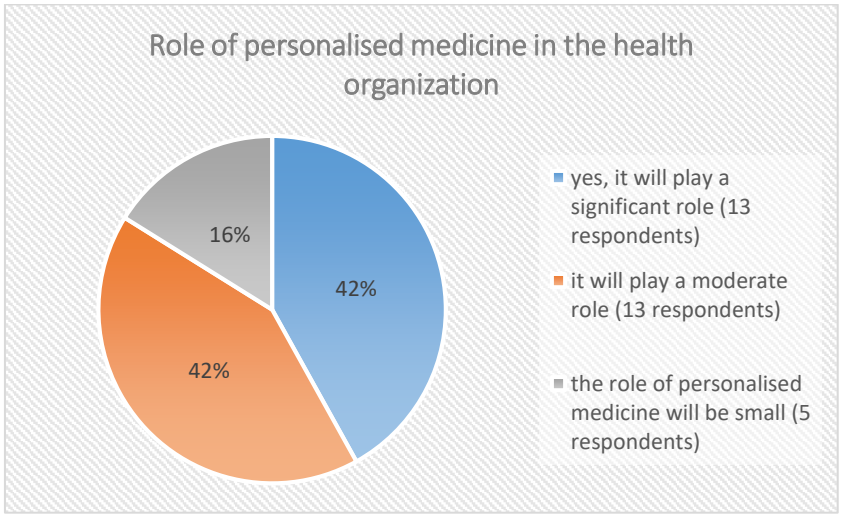

Figure 12. Role of personalised medicine in the organization

For the seamless introduction of personalised medicine and genetics data, it is important, that the integration of genetic data is included into hospital information system or the information system of the health centre. Therefore, it was asked, “whether the

organisation you work for, has plans to integrate patient's genetic data into your hospital/ health centre information system?" The total of 16 respondents (51.61%) replied that "yes, it is planned" and 15 people (48.39%) said that "no, the integration is not planned". 7 cardiologists (58.33%) would answer "yes" and 5 cardiologists (41.67%) would say that "no", the integration is not planned for the genetic data.

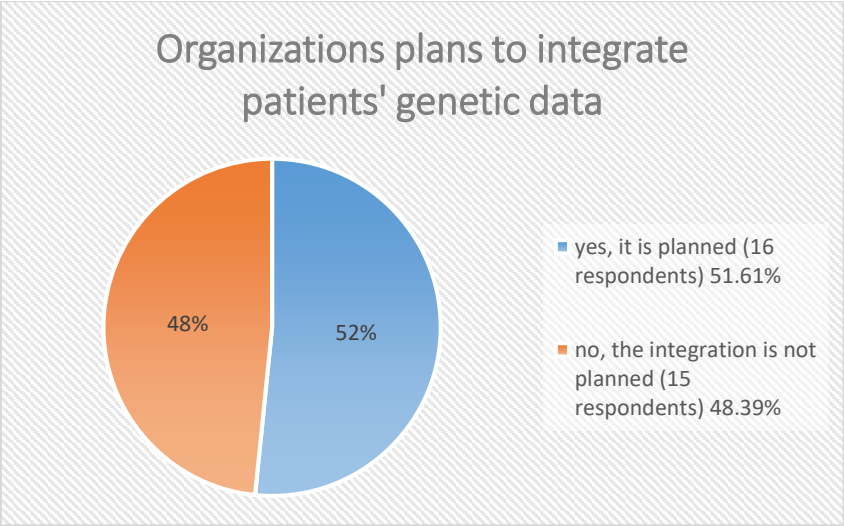

Figure 13. Organization's plans to integrate genetic data

Next step was to understand, whether access to patients' genetic data, would help in CVD or other disease preventive activities? More than an 50% of the total respondents, 17 subjects (54.84%) mentioned that "yes, definitely", the access to patients' genetic data would be useful. 10 respondents (32.26%) mentioned that "maybe", the access to patients' genetic data would be useful addition to the current data access they have. 3 respondents (9.68%) replied that "I am not sure", 1 respondent (3.23%) said that "no, it would not help". In total 27 subjects (87.10%) replied that access to patients' genetic data would be "maybe" or "definitely" useful addition to plan more personalised preventive activities. Form the cardiologists point of view, 7 respondents (58.33%) replied, that "yes, definitely" it would be of importance and 5 respondents (41.67%) would say that "maybe" there is additional value in the genetic information of the patients.

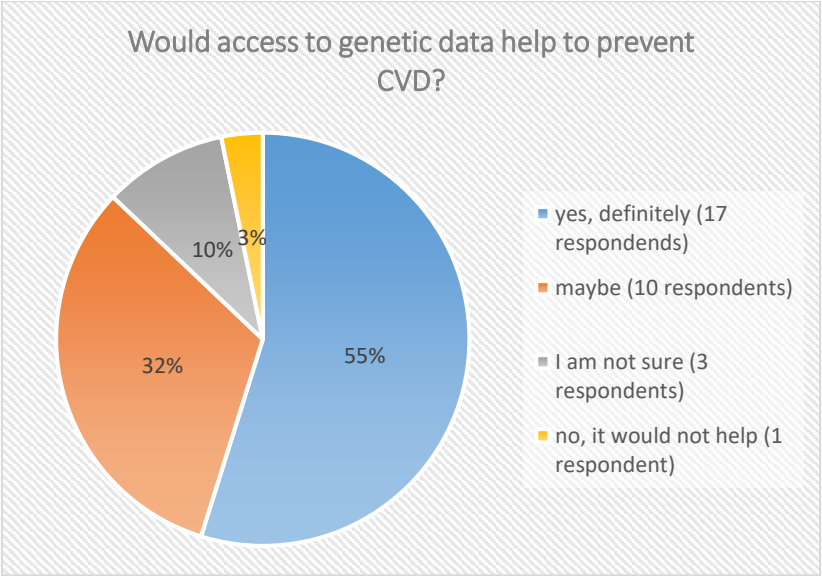

Figure 14.Genetic data access and CVD prevention

As next, it was asked about how comfortable would you feel yourself about consulting the patients according to their genetic data? The reason behind this question was to gain knowledge about the fact, whether the medical personnel would have enough education, knowledge or expertise to consult their patients according to the genetic data they have from the patients. Majority of the respondents 24 people (77.42%) replied that "I don't have enough education or training in that, but I would be willing to learn about it." 2 people (6.45%) replied that "I can handle it" meaning that they are able to understand the genetic information to the extent that they can consult their patients according to that. 4 respondents (12.90%) mentioned that "health coach or someone else should do it" and only 1 respondent (3.23%) replied that "I would not be able to handle it and I am not interested in learning about it."

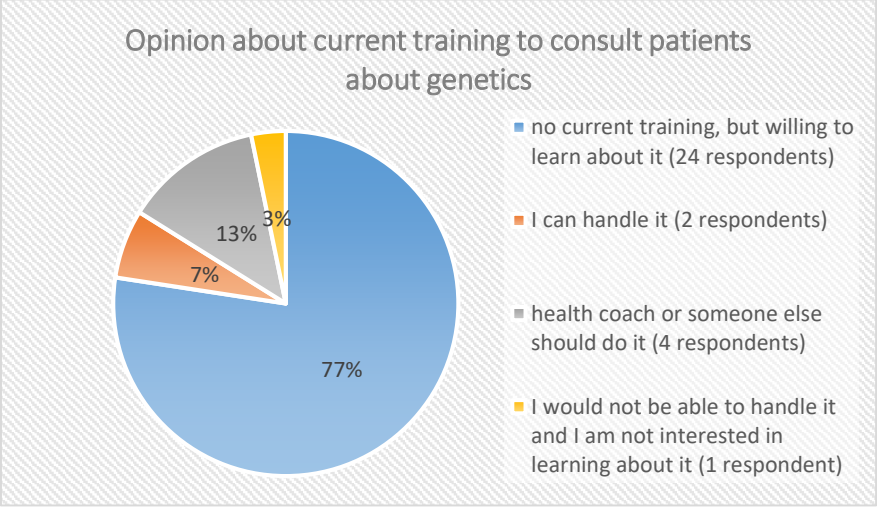

Figure 15. Health professionals' opinion about own genetic testing knowledge

To the question, whether the respondents see additional value in having access to patients' genetic data, the majority or 24 respondents (77.42%) said "yes", they see additional value in having access to patients' genetic data and 7 respondents (22.58%) did not see any value in having access to patients' genetic data. Among the cardiologists the answers would be following: 10 respondents (83.33%) did reply that "yes", there is additional value to having access to patients' genetic data and 2 respondents (16.67%) replied that "no", there is no additional value in them in having access to patients' genetic data.

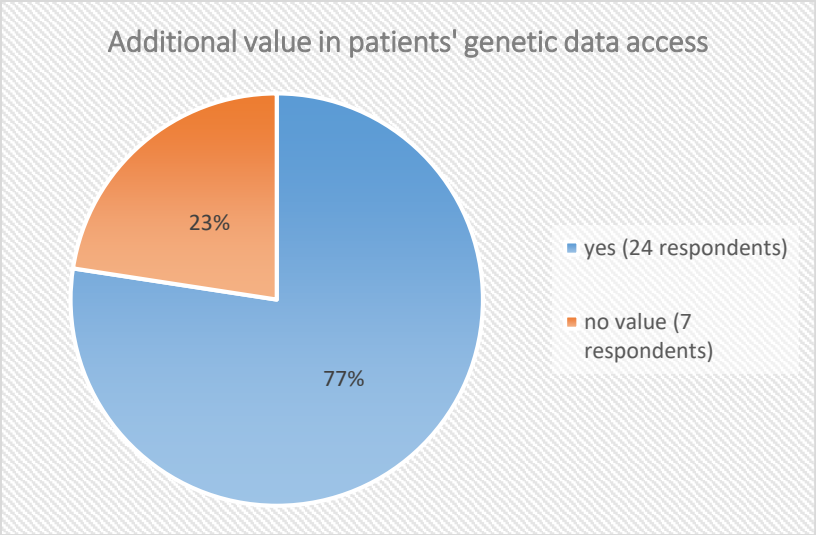

Figure 16. Additional value in patients' genetic data

Since CVD preventive or treatment activities in general may need eventually some additional data entry from the patients into their own activity monitors and/or other devices, it was asked if the data collected by the patients (from activity monitors or other specified devices), when integrated into the hospital/health centre information system would have additional value. 20 respondents (64.52%) replied that "yes", they see additional value in having access to patients own data entered, 10 respondents (32.26%) replied that "difficult to say" and 1 respondent (3.23%) replied that there would be no value in having access to patients own data from their hospital or health centre information system.

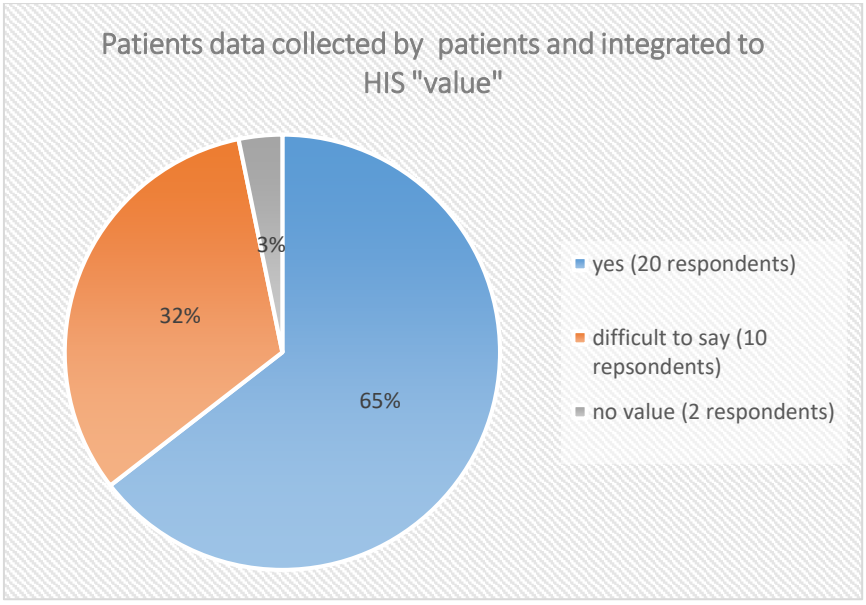

Figure 17. Patients data collected and integrated to HIS

Currently, patients' genetic data is not integrated in the hospital information system or there is no access to it from the patient portal, since that data is not available in the patient portal. Therefore, it was asked, if patients genetic data would be included into the health centre or hospital information system, then in what format would you like to receive access to it? 20 respondents (64.52%) replied that "In a patient summary form, previously analysed by AI or health analyst and sent to me prior to meeting with the patient". 5 respondents (16.13%) replied that "It is sufficient, if the information is available from my organisations information system, I can handle the analysis myself". 1 respondent (3.23%) replied that "I don't see an additional value in that" and 5 respondents (16.13%) replied that "I would not use this information because my profession does not need it (I am a nurse or other medical personnel)". Cardiologists answers for the format of the data was that 11 respondents (91.67%) would like to receive a patient summary, previously analysed by AI or health analyst and presented to them before meeting with the patient. 1 respondent (8.33%) replied that it is sufficient if the genetic data is accessible from the hospital information system.

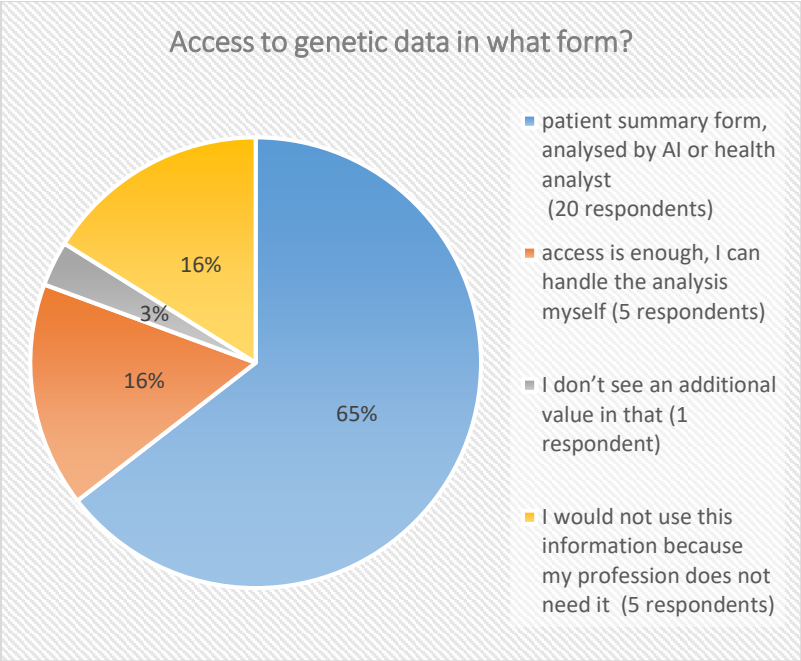

Figure 18. Access to patients' genetic data in what form

In the survey, in the last question, the respondents could add additional comment they had about the state of personalised medicine in Estonia or access to patients' genetic data from the hospital information system or about related topics. The question was not obligatory, and the replies were following: "It is the medicine of the future", "At the moment, there is not enough research done about the patient treatment recommendations according to genetic data and it is difficult to integrate this knowledge into everyday praxis. In the current HIS, there is no integration of patients' priority blood analysis results, which are available in the patient portal, not to mention about genetic data", "The only idea behind personalised medicine is to find a suitable treatment for a concrete person. In other aspects it is recommended to assess the family and make conclusions from there – it is not needed to do expensive studies, if because of which is to conclude that there is no cure for that or you need just general suggestions", "It seems that the topic is very popular among the patients, looking at the topics they bring up when coming to visits. Now there seems to be great interest in becoming a gene donor…" Some respondents did not reply or add any extra comments in this field.

6 Discussion

In the current chapter, the author discusses research outcomes respectively to the predefined research objectives and research questions. Since there were two hypotheses set before the study, the research outcomes were divided into two parts: population opinion and the opinion of health professionals. As research limitations, the author describes possible shortcomings of the study design. Finally, author sets the possible future perspectives.

6.1 Research outcomes: population opinion

In the population opinion and views about personalised medicine and gene testing findings show that the topic of genetic testing and having access to own genetic data is of big importance and study participants were interested in taking part and giving their opinion about the topic. Before interpreting the data of the survey respondents about their opinion on genetic testing, it was first assessed how well do they rate their own health, the health of their family members (mainly their parents and grandparents) and their current physical activity level. The sex of the study participants in the selected group was 55 females (87.30%), and 8 males (12.70%).

From the 63 of total highly educated survey respondents, most of the respondents', 59 people (93.65%) had bachelors or master's degree and from the total number, 55 people (87.3%), rated their health state as "good", "very good" or "excellent". As their current weekly physical activity, as one of the activities to prevent cardiovascular diseases and according to European Guidelines on cardiovascular prevention and other world-wide recommendations to have moderate exercise at least 3 times a week at least total of 90 minutes per week [5], 41 people (65.07%) out of 63 replied that they were doing sports more that 1 – 3 hours and 3+ hours a week, the rest, 22 persons (34.93%) were doing less sports than 1 hour a week. According to the survey results, it could be assumed, that 41 people (65.07%) of the subjects do currently enough weekly physical activities to keep and maintain their heart healthy and 22 people (34.93%) do not have enough weekly physical training and over time could need to modify their lifestyle to add more training to ensure healthy heart condition. This is assumed in case that when selected weekly activities "1 – 3 hrs a week" could be interpreted so that people do exercise at least about

1.5 – 3 hours a week. If in the category "1 – 3 hours a week" some people really did exercise just 1 hour a week, then the total number of currently assumed as "doing enough physical activities" from the 65.07% would be lower.

To understand, what are the channels and from where people currently look for their health information it was asked about their preferences. Answer to the question, from where and how the survey respondents would like to receive information or feedback about their health stat, was not so homogeneous. Only 3 people (4.76%) out of 63 replied that they can handle their health information themselves and do not need any additional help. In the answer choices given there were 2 options about currently available options – receiving the information electronically from the patient portal or in person by their family doctor. Only 5 people (7.94%) replied that they would like to receive their health information from the patient portal and 16 people (25.40%) were interested in receiving their health information personally from their general practitioner (GP). The total percentage of people receiving information from the currently available channels was 21 people (33.34%). The rest 39 people (61.8%) were interested in receiving feedback in some novel ways, which don't exist now.

In the survey, it was not additionally explained, what those new ways exactly were, and the respondents had to guess from the information given to them in the survey questions. From the 39 people (61.8%) choosing from the novel ways of feedback, 6 people (9.52%) were interested in receiving their health information from a personal health application such as My Health Account for example, 12 people (19.05%) would like to have their information analysed by the AI and the summary sent to My Health Account and the total number of 21 people (33.33%) would like to receive feedback from a Health Coach, who would previously analyse their data and give a short summary to the person. It is interesting to find, that above 60% of the respondents choose for the information receiving formats options, that did not exist today yet.

It has been assumed in the hypothesis part, that population is interested and willing in receiving personal health data in more summarized way and previously analysed by AI and explained to them by a Health Coach as an alternative to the family doctor from whom they currently receive their feedback. It is good to see, that the population would not mind receiving information from electronic sources and other than their current GPs, who

currently, according to Centre for Political Studies Praxis research in 2017, do not have enough extra time for the patients to explain any additional topics expect the particular problem or reason patient is coming to see a doctor [40]. In the survey done by Kantar Emor about the population' satisfaction with the primary care physicians and specialists work they find that the patient portal knowledge and importance is high, but highly educated people are still expecting other ways for feedback on their personal health. [41] The results can be interpreted from the fact that the survey subjects were highly educated, 59 people (93.65%) of the total 63 people having at least an undergraduate or graduate degree, being young and most likely used to using other electronic channels every day to handle daily tasks.

For the survey question, whether a person would consider to lose weight when their family doctor would suggest that since it would be beneficial for their health, then 42 people (66.67%) answered that "Yes, I would do it", 20 people (31.75%) answered that "Yes, I would consider it", and only 1 person (1.59%) said that he/she knew the importance of it but would not see the benefit in that. None of the respondents said that they would not do it. To the question, where genetics was involved, and the respondent had to imagine that according to their genetic test results and according to the genetic risk score (GRS) they would belong into the high-risk score class of developing any heart diseases. The question was whether they would then consider to lifestyle change? 44 people (69.84%) answered that yes, they would change their lifestyle to prolong their lives and live a healthier life. 16 people (25.40%) mentioned, that they would "maybe" change their lifestyle to a healthier one. 2 people (3.17%) said that they don't believe in genetics and 1 person (1.59%) said "no" to any changes. The comparison between family doctors influence vs just receiving personal genetic information in some electronic formats shows, that slightly more people do believe more in having their genetic data analysed by AI, health analytics or Health Coach, rather than their family doctor (FD) – 44 people (69.84%) vs 42 people (66.67%).

When there was the question where genetics was involved, and the survey respondents replied that they would consider lifestyle changes "maybe", it would need more clarification and understanding, what is needed to be more confident about genetic results and be clearer in question replies. It could also be interpreted, that there is more information needed to make such decisions. However, the influence and trust in family

doctors is high among the survey respondents, which means that when the FD would consult according to the patients' genetic data, then the patients would more likely accept their opinion and guidance.

All in all, as a result of current research we can conclude that according to the current study, population is ready and willing to use the advantages of the personalised medicine. Hypothesis which was set at the beginning of the research was that population is ready to have genetic data included into the prevention process of CVD and is expecting to have primary care level consultations according the personalised medicine point of view. Population is ready to accept novel information receiving formats - health summary by the health coach or health information analysed by AI.

In conclusion, the comparison between family doctors influence when giving feedback and recommendations about preventive CVD measures vs just receiving personal genetic information in some electronic formats shows, that slightly more people do believe more in having their genetic data analysed by AI, health analytics or Health Coach, 44 people (69.84%) rather than their GP 42 people (66.67%). In addition, we can clearly say that population is expecting novel information exchange formats. The total percentage of people who would like to receive information from the currently available channels (GP or patient portal) was 21 people (33.34%), and the majority, 39 people (61.8%) were interested in receiving feedback in some novel ways, such as having data directly transferred in analysed way to My Health Account, receiving information and consultation from the Health Coach.

6.2 Research outcomes: health professionals opinions

Findings in health professionals survey show that health professionals were mostly accepting the ideas of personalised medicine, would like to have genetic testing results integrated into the hospital/ health centre information system and would benefit from having access to patients' genetic data.

In total, 30 respondents (99%) regard the personalised medicine as "important", "very important" or "somewhat important". Above 80% of the respondents, 26 people (83.88%) reply that in the next 5 years, the personalised medicine would play "an important role"

or “a moderate role” in their organisation. Integration of the genetic data is planned into the hospital or medical centre information systems in the organisations of 16 respondents (51.61%). 24 of the respondents (77.42%) see additional value in having access to the patients’ genetic data. The low percentage of respondents and low trust in having genetic data integrated into their HIS may be interpreted from the fact that there are currently other more important issues do be dealt with such as for example priority blood analysis from patient portal data, which is not integrated in the HIS yet as brought out in the comments of health professionals in the survey. If the health professionals would have access to patients’ genetic data, 17 respondents (54.84%) say that “yes, definitely”, it would help them to plan the preventive activities for the patients. A high percentage of 32.26% (10 respondents) replied that “maybe” the access to the patients’ genetic data would be of an additional value. This number shows, that it may be currently still a little unclear to the health professionals, what is the specific value they would have from having access to patients’ genetic data, how the information accessed is accessed and from where and in what format the information is presented to the health professionals. There could also be a speculation that changes in health professionals everyday working praxis are not something everybody is looking for and changes may require time to cope with.

Moreover, in terms of consulting the patient according to the personal genetic data, surprisingly 24 respondents (77.42%) replied that they don’t have enough training or education to consult their patients about their genetic risks or according to their genetic data, but they would be willing to learn about it. 2 people (6.45%) declared themselves as having enough training and knowledge in genetics and can consult their patients according to that. A significant number of respondents, 4 people (12.90%) replied that a Health Coach or someone else should do the patient consulting regarding the genetic data. It is interesting, that this answer was chosen, since there is currently no such medical profession existing and there are currently no Health Coaches in Estonia and the tasks and responsibilities of a Health Coach has not been defined yet. Who exactly would be a health coach, who could train them, how long would the training be, what would be the subjects the health coach needs to be trained in and so on – this needs to be defined over time.

All in all, health professionals are willing to work with the patients’ genetic data and they would like to have as compact and clear access to it as possible. Survey results show that

20 respondents (64.52%) would like to have a patient summary presented before meeting the patient, which ideally would be previously analysed by AI or a Health Analyst. Here is seen the importance in developing a patient summary or something similar, which would make the everyday work of medical personnel working with CVD preventive activities. 5 respondents (16.13%) replied that the patients' genetic data could be accessible from the HIS, which means that the importance of integrating the genetic data into is significant.

In the hypotheses at the beginning of the theses was stated that health professionals are willing to use the benefits of personalised medicine and accepting genetic data for broader patient health information availability and they are ready to accept novel information receiving formats for example patient summary analysed by AI or by the health coach. According to that, we can conclude that the survey results meet the hypothesis and the hypothesis is positive.

As a total conclusion from population views and from the health professionals' responses, it is evident, that both parties would accept the idea of including personal genetic test results into the current CVD preventive activities. Both respondents did favour the patient summary, or the information presented to them in a summarized format. The health professionals as well as the population did like the idea of artificial intelligence analysing the data and summarizing the important parts for the receiving party (whether health professionals or population). The need for health analyst and health analytics was brought up in both survey replies, even though this position does not exist yet and the survey respondents may not know for sure, what exactly would the person do, what his or her qualification would be and what would he/she be capable of doing.

6.3 Proposals for change in the CVD prevention according to PM

Changes for the health professionals

Since health professionals have the clear interest in having access to patients' data in as simple and clear ways as possible, then following changes are proposed by the author.

- Patient summary and health analytics

Heath professionals mentioned in their replies in the survey conducted, that they would like to receive a summary of the current state of patients' data. Health professionals would

like to receive a patient summary about patients' health state. Therefore, a patient summary should be composed by AI or health analyst so that the health professionals don't have to go through all patients' available data in different systems but just receive a summary of most critical data.

- Integration

If changes are to be made, then they should be integrated with the existing information systems (whether with hospital information system or GP's praxis information system), otherwise it is something difficult to use and not practical. Therefore, genetic data to be integrated into HIS or patient portal as well.

Changes for the population (*patient empowerment*)

According to the study results, we can clearly see, that there is a definite need and readiness from the population side for patient involvement in their treatment and health consultations. Therefore, following changes will be proposed in CVD prevention.

- Ways of communication – Health Coach

The necessity and usefulness of such a profession as a health coach was mentioned in both survey results – health professionals and population survey. The additional value is being seen in having health data explained to the person in an understandable way so that the person can make informed decisions about his/her health.

- Patients' access to their health data – My Health account

Population has mentioned the importance of having real time access to their own health data and health state, therefore, an account where all patients' relevant health data should be available and accessible is desired.

6.4 Research Limitations

The study had a relatively small sample population. Therefore, further investigations to evaluate the acceptance of population and the health professionals about the genetic testing should be carried out in Estonia. In addition, the population sample could be enlarged via adding the international scope to the study, since the topic of genetic testing and whether the GRS studies are enough to study and to carry out to improve the current CVD prevention activities from the personalised medicine point of view.

Even though the focus of this study was to survey younger, highly educated people, it could be of advantage to research other age groups and people with a different educational level, to receive a broader overview of the population opinion. From the health professionals side, the concentration only on cardiologists and family doctors would be proposed, as they are currently the main contact persons for patients in CVD preventive or treatment activities and plans.

As in the results outcome already mentioned, some survey questions could have been more formed differently or be little more specified. For example the duration of activity 1 – 3 hours a week should have been 1,5 – 3 hours a week, since the recommended minimum weekly physical activity by World Health Organisation [1] and 2016 European Guidelines on cardiovascular diseases prevention in clinical practice [5] recommend at least 30 minutes of active physical activities at least 3 – 5 times a week. In the conducted survey, there is a question for health professionals about patients' activity monitor data, which patients are filling out themselves and whether the data should be available to the health professionals from HIS or patient portal. The answer to this question and the question itself is relevant, when we talk about the CVD treatment or prevention activities, where it is important to measure the exact amount of personal physical activities and the data should be available to the treating physician or FD in the most convenient way. In this study, this question is not relevant, since the focus of the study was to assess the genetic risk scores and the population acceptance on genetic testing.

Even though the research results confirmed the hypothesis set at the beginning of the thesis, that population as well as the health professionals would like to receive a summary of their health state or patient summary respectively, it could be researched in more detail, what would be exact format of it, who and how will it be composed, who would analyse it and in what format would it be available to the interested parties.

Moreover, the concept of Health Coach could be researched in more detail. As the study results showed, the idea of receiving summarised information about personal health state (population view) or having patients genetic data analysed (health professionals view) by a Health Coach, was favourable by both – population and the health professionals.

In conclusion, the abovementioned additions could increase the relevance of the findings, give deeper understanding of the topic of genetic testing and GRS importance in CVD prevention.

6.5 Future perspectives

The future research on CVD prevention through personalised medicine approach could go deeper into the investigation of following topics:

- health analytics or artificial intelligence (AI)

It could be researched how health analytics and AI can be of assistance to speed up the processes so that the parties involved would be receiving the genetics data analysed for them in the appropriate and understandable way.

- patient summary (for health professionals) or health state summary (for people)

The need to define and specify what exactly constitutes a summary, how and from whom it will be composed, and how and from where it is accessible.

- Health Coach

The importance and need to define the nature and responsibilities of a health coach, how his/her services are accessed, promoted and carried out. Would the Health Coach work independently and offer the services as an agency or would the service operate under a family doctors praxis or another medical institution or hospital. Would the service be paid by health insurance fund as a preventive CVD measure?

7 Summary

Cardiovascular diseases are the main cause of death in Estonia and worldwide and it has a negative socio economic impact because mostly, people in the working age develop CVD [1]. The disease itself, the onset age of it and the on-time treatment can make the burden to the whole society and therefore, its early detection, preventive activities and treatment could be the main indicators improving the amount of burden to the society. Public support will be crucial to realize health improvements and if researchers can engage the public in a regular, transparent dialogue, then this could contribute to greater understanding of the research processes, the design of efficient and effective genetic health services, informed by the public who would use them [22]. Because of that, the alternative and novel ways of detecting CVD as early as possible as well as the population and health care professionals' acceptance about including genetic testing result to the CVD preventive measurements is highly necessary.

The aim of the master thesis was to assess the acceptance and understanding of the genetic information by the population and health professionals. Therefore, two surveys were composed, and the research conducted to receive broader understanding of the current state of the population and health professionals' views on the topic.

The main outcomes and conclusions of the thesis are following:

- In the population results: The comparison between family doctors influence when giving feedback and recommendations about preventive CVD measures vs just receiving personal genetic information in some electronic formats shows, that slightly more people do believe more in having their genetic data analysed by AI, health analytics or Health Coach, 44 people (69.84%) rather than their GP 42 people (66.67%). In addition, we can clearly say that population is expecting novel information exchange formats. The total percentage of people who would like to receive information from the currently available channels (GP or patient portal) was 21 people (33.34%), and the majority, 39 people (61.8%) were interested in receiving feedback in some novel ways, such as having data directly transferred

in analysed way to My Health Account, receiving information and consultation from the Health Coach.

- In the health professionals' results: All in all, health professionals are willing to work with the patients' genetic data and they would like to have as compact and clear access to it as possible. Survey results show that 20 respondents (64.52%) would like to have a patient summary presented before meeting the patient, which ideally would be previously analysed by AI or a Health Analyst. Here is seen the importance in developing a patient summary or something similar, which would make the everyday work of medical personnel working with CVD preventive activities. 5 respondents (16.13%) replied that the patients' genetic data could be accessible from the HIS, which means that the importance of integrating the genetic data into is significant.

Therefore, populations' genetic testing should be continued and included in the CVD prevention process to identify as early as possible the persons who need the immediate treatment or lifestyle changes. Currently, genetic testing data results are not integrated into any information systems (such as HIS, information system in the GP office or the Estonian state patient portal) where health professionals or population would have easy access to it. The summary of the patient health state, e.g. "*patient summary*" for the health professionals or "*my health summary*" for the general population and there is currently no specific person or electronic system, or portal assigned where genetic data would currently be analysed, and the outcomes summarised. Therefore, the need for a Health Coach, inclusion of AI and health analytics is crucial for the success of integration of patients genetic for CVD prevention process.

References

[1] WHO, “WHO _ Cardiovascular diseases (CVDs),” *Cardiovascular diseases (CVDs)*. 2015.

[2] R. Baetta, M. Pontremoli, A. Martinez Fernandez, C. M. Spickett, and C. Banfi, “Proteomics in cardiovascular diseases: Unveiling sex and gender differences in the era of precision medicine,” *J. Proteomics*, vol. 173, pp. 62–76, 2018.

[3] Eurostat, “Causes of death statistics - Statistics Explained,” *Eurostat Stat. Explain.*, 2017.

[4] M. Kaldmäe, G. Zemtsovskaja, J. Abina, T. Land, and M. Viigimaa, “Prevalence of cardiovascular disease risk factors in Tallinn, Estonia,” *Med.*, vol. 53, no. 4, 2017.

[5] M. F. Piepoli, “2016 European Guidelines on cardiovascular disease prevention in clinical practice,” *Int. J. Behav. Med.*, vol. 24, no. 3, pp. 321–419, 2017.

[6] M. Nichols *et al.*, *European cardiovascular disease statistics 2012*. 2012.

[7] GBD 2016 Causes of Death Collaborators, “Global, regional, and national age-sex specific mortality for 264 causes of death, 1980–2016: a systematic analysis for the Global Burden of Disease Study 2016,” *Lancet*, vol. 390, no. 10100, pp. 1151–1210, 2017.

[8] G. Danaei, E. B. Rimm, S. Oza, S. C. Kulkarni, C. J. L. Murray, and M. Ezzati, “The promise of prevention: The effects of four preventable risk factors on national life expectancy and life expectancy disparities by race and county in the United States,” *PLoS Med.*, vol. 7, no. 3, pp. 1–13, 2010.

[9] K. K. Jain, “A critical review of the Royal Society’s report on personalized medicine,” *Drug Discovery Today*, vol. 11, no. 13–14, pp. 573–575, 2006.

[10] J. Stewart, G. Manmathan, and P. Wilkinson, “Primary prevention of cardiovascular disease: A review of contemporary guidance and literature,” *JRSM Cardiovasc. Dis.*, vol. 6, p. 204800401668721, 2017.

[11] C. C. Imes and F. M. Lewis, “Family history of cardiovascular disease, perceived cardiovascular disease risk, and health-related behavior: a review of the literature.,” *J. Cardiovasc. Nurs.*, vol. 29, no. 2, pp. 108–29, 2014.

[12] P. C. Kollia N, Panagiotakos DB, Chrysohoou C, Georgousopoulou E, Tousoulis D, Stefanadis C, Papageorgiou C, “Determinants of healthy ageing and its relation to 10-year cardiovascular disease incidence: the ATTICA study,” *Cent Eur J Public Heal.*, 2018.

[13] S. Kathiresan and D. Srivastava, “Genetics of human cardiovascular disease,” *Cell*, vol. 148, no. 6. pp. 1242–1257, 2012.

[14] S. K. Nadar and K. Sandhu, “Genes and cardiovascular disease: Where do we go from here?,” *Sultan Qaboos University Medical Journal*, vol. 15, no. 4, pp. e448–e451, 2015.

[15] A. Singleton, L. H. Erby, K. V. Foisie, and K. A. Kaphingst, “Informed choice in direct-to-consumer genetic testing (DTCGT) websites: A content analysis of benefits, risks, and limitations,” *J. Genet. Couns.*, vol. 21, no. 3, pp. 433–439, 2012.

[16] M. Paquette, M. Chong, S. Thériault, R. Dufour, G. Paré, and A. Baass, “Polygenic risk score predicts prevalence of cardiovascular disease in patients with familial hypercholesterolemia,” *J. Clin. Lipidol.*, vol. 11, no. 3, p. 725–732.e5, 2017.

[17] H. Tada *et al.*, “Twelve-single nucleotide polymorphism genetic risk score identifies individuals at increased risk for future atrial fibrillation and stroke,” *Stroke*, vol. 45, no. 10, pp. 2856–2862, 2014.

[18] K. K. Jain, “Personalized Management of Cardiovascular Disorders,” *Medical Principles and Practice*, vol. 26, no. 5. pp. 399–414, 2017.

[19] J. W. Knowles *et al.*, “Impact of a Genetic Risk Score for Coronary Artery Disease on Reducing Cardiovascular Risk: A Pilot Randomized Controlled Study,” *Front. Cardiovasc. Med.*, vol. 4, 2017.

[20] S. Schee genannt Halfmann, N. Evangelatos, P. Schröder-Bäck, and A. Brand, “European healthcare systems readiness to shift from ‘one-size fits all’ to personalized medicine,” *Per. Med.*, vol. 14, no. 1, pp. 63–74, 2017.

[21] D. Horgan *et al.*, “Is precision medicine the route to a healthy world?,” *The Lancet*, vol. 386, no. 9991. pp. 336–337, 2015.

[22] H. Etchegary, J. Green, P. Parfrey, C. Street, and D. Pullman, “Community engagement with genetics: Public perceptions and expectations about genetics research,” *Heal. Expect.*, vol. 18, no. 5, pp. 1413–1425, 2015.

[23] J. E. Ostergren *et al.*, “How Well Do Customers of Direct-to-Consumer Personal Genomic Testing Services Comprehend Genetic Test Results? Findings from the Impact of Personal Genomics Study for the PGen Study Group,” *Public Health Genomics*, vol. 18, no. 4, pp. 216–224, 2015.

[24] D. A. Marshall *et al.*, “The price of whole-genome sequencing may be decreasing, but who will be sequenced?,” *Per. Med.*, vol. 14, no. 3, pp. 203–211, 2017.

[25] I. H. Lee, H. Y. Kang, H. S. Suh, S. Lee, E. S. Oh, and H. Jeong, “Awareness and attitude of the public toward personalized medicine in Korea,” *PLoS One*, vol. 13, no. 2, 2018.

[26] M. Gastrow, B. Roberts, V. Reddy, and S. Ismail, “Public perceptions of biotechnology in South Africa,” *S. Afr. J. Sci.*, vol. 114, no. 1–2, 2018.

[27] P. Chow-White, D. Ha, and J. Laskin, “Knowledge, attitudes, and values among physicians working with clinical genomics: A survey of medical oncologists,” *Hum. Resour. Health*, vol. 15, no. 1, 2017.

[28] Wikipedia, “Personalized Medicine,” 2018.

[29] K. Kichko, P. Marschall, and S. Flessa, “Personalized medicine in the U.S. and

Germany: Awareness, acceptance, use and preconditions for the wide implementation into the medical standard," *J. Pers. Med.*, vol. 6, no. 2, 2016.

[30] L. Leitsalu and A. Metspalu, "From Biobanking to Precision Medicine: The Estonian Experience," in *Genomic and Precision Medicine: Foundations, Translation, and Implementation: Third Edition*, 2016, pp. 119–129.

[31] A. Metspalu, "About Estonian Genome Center, University of Tartu (The Estonian Biobank)." [Online]. Available: https://www.geenivaramu.ee/en/about-us. [Accessed: 30-Apr-2018].

[32] S. Wunnenburger *et al.*, "Associations between genetic risk variants for kidney diseases and kidney disease etiology," *Sci. Rep.*, vol. 7, no. 1, 2017.

[33] T. M. Rutten-Jacobs LCA, Tozer DJ, Duering M, Malik R, Dichgans M, Markus HS, "Genetic Study of White Matter Integrity in UK Biobank (N=8448) and the Overlap With Stroke, Depression, and Dementia.," *PubMed*, 2018.

[34] C. W. You L, Li C, Zhao J, Wang DW, "Associations of common variants at ALDH2 gene and the risk of stroke in patients with coronary artery diseases undergoing percutaneous coronary intervention.," *PubMed*, 2018.

[35] B. Bruce and J. F. Fries, "The Stanford Health Assessment Questionnaire: Dimensions and practical applications," *Health and Quality of Life Outcomes*, vol. 1. 2003.

[36] R. S. Safeer, C. E. Cooke, and J. Keenan, "The impact of health literacy on cardiovascular disease," *Vascular Health and Risk Management*, vol. 2, no. 4. pp. 457–464, 2006.

[37] L. Cusack, C. B. Del Mar, I. Chalmers, and T. C. Hoffmann, "Educational interventions to improve people's understanding of key concepts in assessing the effects of health interventions: A systematic review protocol," *Syst. Rev.*, vol. 5, no. 1, 2016.

[38] C. W. Cheng YL, Shu JH, Hsu HC, Liang Y, Chou RH, Hsu PF1, Wang YJ1, Ding YZ, Liou TL, Wang YW1, Huang SS1, Lin CC1, Lu TM1,4,5, Leu

HB1,4,5, Lin SJ1, “High health literacy is associated with less obesity and lower Framingham risk score: Sub-study of the VGH-HEALTHCARE trial.,” 2018.

[39] S. Crengle *et al.*, “Effect of a health literacy intervention trial on knowledge about cardiovascular disease medications among Indigenous peoples in Australia, Canada and New Zealand,” *BMJ Open*, vol. 8, no. 1, p. e018569, 2018.

[40] K. K. Laura Aaben, Gerli Paat-Ahi, Ülla-Karin Nurm, *Rahvastiku tervise arengukava 2009-2020 vahehindamine. Südame- ja veresoonkonnahaiguste valdkonna aruanne*. Poliitikauuringute Keskus Praxis, 2017.

[41] Kantar Emor, “Eesti elanike hinnangul on arstiabi kvaliteet hea, muret teeb arstiabi kättesaadavus,” 2016. [Online]. Available: http://www.sm.ee/et/uudised/uuring-eesti-elanike-hinnangul-arstiabi-kvaliteet-hea-muret-teeb-arstiabi-kattesaadavus. [Accessed: 10-Apr-2018].

Appendix 1 – Survey questions for population

Background

1. Age (18-20, 21-30, 31-40, 41-50, 51-60, 61-70, 71-80)
2. Sex (male, female)
3. Education
    a. Primary
    b. Secondary
    c. University
    d. Postgraduate

General health

4. Are you aware of if you have any diseases related to cardiovascular diseases (high blood pressure, obesity, hypertension, etc.)
    a. Yes
    b. No
    c. Don’t know
5. Has any of your family members (parents or grandparents) been diagnosed with Diabetes 1 or Diabetes 2?
    a. Yes
    b. No
    c. Don’t know
6. In general, would you say your health is:
    a. Excellent
    b. Very good
    c. Good
    d. Fair
    e. Poor

Physical activity

7. During the past week, even if it was not a typical week for you, how much total time (for the entire week) did you spend on each of the following activities (walking, swimming, cycling, etc.).
    a. 0 minutes
    b. Less than 30 minutes a week

c. 30-60 minutes a week
d. 1-3 hours a week
e. 3 hours and more per week

Access to your health data

8. How would you like to get an opinion about your current health state?
    a. From the patient portal, analysing the data myself
    b. From my personal health app (e.g. My Health account)
    c. My family doctor does give me sufficient feedback in person
    d. I wouldn't mind having my data analysed by AI and receiving a short health summary
    e. From the personal Health Coach who would analyse my data and give me feedback (via online or in person)
    f. I can handle my health myself
9. If your doctor would tell you to lose weight for health benefit, would you do it?
    a. Yes, I would do it
    b. Yes, I would consider it
    c. Not sure, if it is important
    d. Definitely no
10. Studies show that 30% of our health is influenced by our genes. If you would get to know (through gene testing) that you belong to the high-risk behaviour class in developing any heart diseases. Would you then consider changing your lifestyle to prevent the onset of a disease or gain few life years?
    a. Yes, definitely
    b. No
    c. Maybe
    d. I don’t believe in genetics

Appendix 2 – Survey questions for health professionals

1. What is your occupation?
    a. Medical doctor (cardiology)
    b. Medical doctor (other)
    c. Nurse
    d. Other supporting hospital personnel
2. What is your opinion about precision medicine (personalised medicine)?
    a. Is needed
    b. Not needed
    c. Very useful
    d. Slightly useful
3. Would precision medicine play a significant role in your organizations in the next five years?
    a. A significant role
    b. An average role
    c. A small role
    d. No role
4. Does your organization have plans to integrate Genomics into its Electronic Health record?
    a. Yes
    b. No
5. If you would have access to patient's genetic data, would it help you in preventing CVD in patients?
    a. Definitely, yes
    b. Yes
    c. Maybe
    d. No, it will not help
    e. Not sure
6. Are you comfortable and do you have sufficient education/training about discussing and consulting genetic data with your patient?
    a. Yes, I can manage it
    b. No, I don't have enough training about it, but I would be willing to learn

    c. No, I can’t manage it and I am not interested in learning about it
    d. A health coach or someone else should do it
7. Do you see value in having access to patients’ genetic data?
    a. Yes
    b. No
8. Do you see value in data, added and collected personally by the patients (such as information from activity monitors about training, sleep, food intake, weight, blood pressure measured regularly at home) in prevention of CVD?
    a. Yes
    b. No
    c. Difficult to say
9. If genetic data is included in Electronic Health record, in what form would you like to receive it?
    a. In a form of patient summary, composed before meeting the patient and analysed by AI or health analyst
    b. Just included in Patient Portal, I can handle the analysis myself
    c. I don’t see any value in that information
    d. I would not use it due to my occupation (e.g. I am a nurse)
10. Anything else you would like to add about personalised (precision) medicine, access to patient’s genetic data or data added by the patient?
    Free text field would be here