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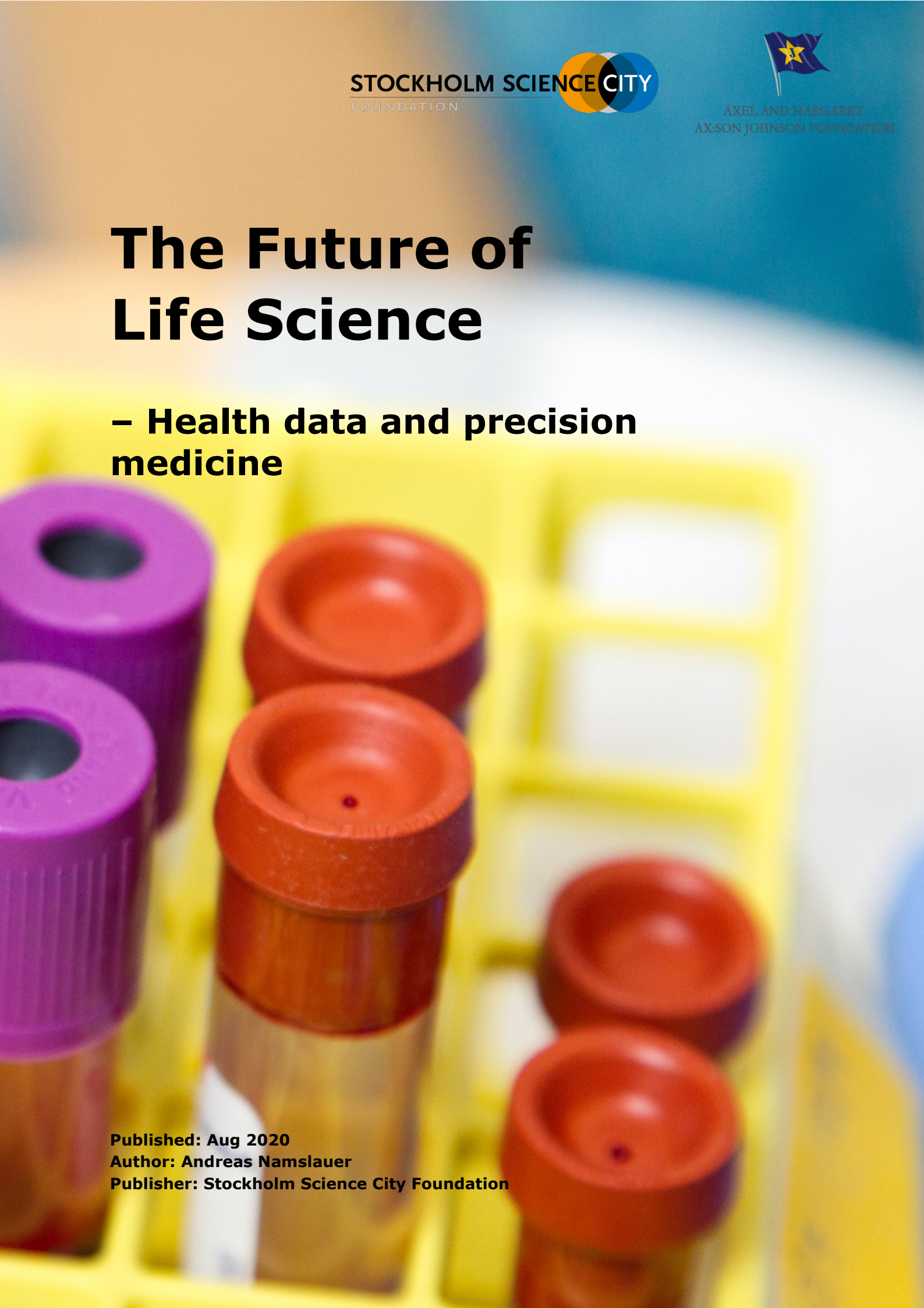
The Future of Life Science

– Health data and precision medicine



Published: Aug 2020
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Publisher: Stockholm Science City Foundation

Summary

Where is life science heading in the future? Life science has always been about deciphering biological complexity and two factors that drive the development in life science are the rapid advances in genetic sequencing and data analytics. In this report, we discuss the benefits, possibilities and risks of using health data in new ways to improve our health, medical treatments and the way healthcare is carried out.

With new technology comes new opportunities to diagnose diseases and to tailor treatments, but also to better predict risks and work preventative. Globally, large investments are made in *precision medicine* with the aim to use genetic data, biological markers and other health data to match patients with treatments that are likely to work. For some cancer types and a few rare diseases, the new processes have already changed outcomes profoundly. The rapid development is powered by increasing international collaborations among researchers, but different national regulations are affecting how patient data can be used for research and how precision medicine can be applied in healthcare.

The national data protection regulations in Sweden have been rather restrictive in an international comparison. Several countries have modified their legislation regarding health data to improve correlations with modern technologies and digital information handling as used today. Their updated legislation enables for development of new therapies as well as other types of innovation. Several stakeholders, including caregivers, healthcare professionals and researchers have pointed out that the Swedish legislation is difficult to interpret and that it hampers the benefits of sharing data. At the same time, the Swedish national life science strategy points out that better utilization of healthcare data is a prerequisite for Sweden to be able to take lead in life science.

The Swedish legislation regarding the storage of information in biobanks and the exchange of patient data between caregivers is currently the subject of government enquiries. If Sweden aims to use healthcare data and data from e.g. genetic screening with the aim to improve healthcare and accelerating research and development of new therapies, the legal framework needs to be modernised in a way that balances benefits for individuals and society with personal integrity and the risk of misuse.

INTRODUCTION

Life science has always been about deciphering biological complexity. With the tools at hand during the 20th century, biomedical scientists had to use a reductionist approach, where problems were studied one at a time. With the rapid development over recent decades of new technology and novel methods of analysing data, we are now – in the 21st century – in a position where complex integrated biological systems can be studied holistically. Two factors that drive this development are the rapid advances in genetic sequencing and data analytics.

Two decades ago, shortly before the Human Genome Project was about to complete the first human genome after 13 years of intense work, Francis Collins, who was then director of the National Center for Human Genome Research, made predictions about the health benefits to be gained from the Human Genome Project. Collins' fundamental idea was that the technology and insights of the Human Genome Project would demonstrate tight causal links between variations in DNA sequences and complex human traits, including the disorders that dominate human illness and death. The results of the Human Genome Project were predicted to transform medical care (by the year 2010), evoke behaviour changes in genetically at-risk individuals, generate the development of new drugs, and improve the effectiveness of old drugs by matching them to patients' genetic backgrounds. This could be seen as an early manifestation of what we now define as precision medicine and it has determined the direction of a large proportion of medical research ever since.

Large projects that have mapped the building blocks of life, such as the Human Genome Project and the Human Protein Atlas, have provided a fundamental basis for understanding life but have also contributed to the progression of technology development. New technology has given us faster, cheaper sequencing techniques, increased computational power, advances in big data analysis and higher resolution imaging capacity. This has allowed an unpreceded increase in our understanding of human biology over the last few decades.

Where we live, our genetic makeup, our family medical history and our lifestyle choices all contribute to our health and well-being. By taking these factors into account, tailored treatments can now be offered for some indications. Precision medicine is an emerging

field that aims to individualise medical treatment. The progression of precision medicine is fundamentally dependent on the collection, handling and analysis of data. The methods used to collect, handle and analyse these data will determine how we can benefit from precision medicine. In this report, we will discuss the benefits, possibilities and risks of using health data in new ways to improve our health, medical treatments and the way healthcare is carried out. What is currently possible? Should large-scale genetic screening be carried out routinely to assess individual risks of disease and enable preventive action? What are the challenges associated with implementing precision medicine in Sweden today? How do we balance personal integrity with benefits for society and improved health for individuals?

This report builds on some of the presentations and discussions at the symposium "Where is life science heading in the future?", with the theme *"Technology-driven science and healthcare - New opportunities and potential risks",* held at Engelsberg Ironworks (*Engelsberg Bruk*), Sweden, in May 2019. It is not the intention of the report to cover all the topics discussed at the symposium. The aim is rather to discuss the possibility of using health data for precision medicine, which was one of the topics of the symposium, covering the status of the field in Sweden and elsewhere. The national data protection regulations in Sweden are rather restrictive compared to those in some other countries on how health data can be used by caregivers and researchers. Some other countries are now rapidly modernizing their legislation to increase access to health data for research and development. Sweden is lagging behind in this field and risks missing out on both related investments and the best utilization of the resources in healthcare. Hence, the authors of this report wish to highlight both the benefits and the risks of using genetic and other individual data in healthcare, research and innovation, as a basis for further discussion. Questions related to health data will be further highlighted at the upcoming symposium in 2021.

The symposia are part of the initiative "The future of Life Science", which is a collaboration between the Axel and Margaret Ax:son Johnson Foundation and the Stockholm Science City Foundation. The aim of this collaboration is to explore how breakthroughs within life science affects the society, healthcare and our lives. The symposia address relevant questions and aim to trigger discussions among experts and researchers from different disciplines, policymakers and executives from industry.

WHAT IS PRECISION MEDICINE AND HOW IS IT USED TODAY?

Our health and wellbeing depend on a broad array of factors such as our genetic makeup, the environment we live in and our lifestyle. In precision medicine, the study of our genes is used alongside information about our lifestyles and environments to help predict which treatments and prevention strategies might work best in a specific group of people.

There is no single definition of the concept of precision medicine, but one that has gained strong support is that precision medicine is the categorisation of patients into groups based on biological information and biomarkers at the molecular level using the fields of genomics, proteomics, metabolomics, epigenomics and pharmacogenomics, thus allowing a patient to be matched with a treatment that is likely to work. This is in contrast to the one-size-fits-all approach, in which disease treatment and prevention strategies are developed for the average person, with less consideration of the differences between individuals. The goal of improving clinical outcomes for individual patients and minimizing unnecessary side effects is inherent in the concept of precision medicine.[1]

So called biomarkers (or biological markers) are crucial when tailoring treatments. A biomarker is a measurable indicator of a biological state or condition. Biomarkers are often measured and evaluated to examine normal biological processes, disease processes, or responses to a therapy.[2] Examples of biomarkers include everything from pulse and blood pressure to specific substances in biological tissues, measurement of the latter sometimes requiring complex laboratory tests. During the last decade, specific variations in our genetic code have become increasingly important as biomarkers.

To date, the lion's share of currently implemented precision medicine processes in healthcare is related to the use of genetic tests to determine which type of cancer or hereditary disease a patient carries with the aim of using the best possible treatments for that individual.

[1] FACT SHEET: President Obama's Precision Medicine Initiative, 2015 https://obamawhitehouse.archives.gov/the-press-office/2015/01/30/fact-sheet-president-obama-s-precision-medicine-initiative
[2] Clin Pharmacol Ther. 2001 Mar;69(3):89-95 https://www.ncbi.nlm.nih.gov/pubmed/11240971

THE PHARMACEUTICAL INDUSTRY IS PROMOTING THE PRECISION MEDICINE CONCEPT

The precision medicine concept has been widely adopted by the pharmaceutical industry. The Personalized Medicine Coalition (PMC) defines personalized/precision medicines as "those therapeutic products for which the label includes reference to specific biological markers, identified by diagnostic tools, that help guide decisions and/or procedures for their use in individual patients." Using this definition, 25 of the 59 new drugs (or entities including drugs, agents and therapeutic biologicals) approved by the U.S. Food and Drug Administration (FDA) in 2018 fell into this category.[3]

The focus for the pharmaceutical industry precision medicine investments is on cancers and rare diseases, where detection of specific biomarkers (often including gene variations) has been developed for use in combination with specific drugs. For example, Roche has developed a so-called "broad companion diagnostic" (CDx) service for genetic analysis of solid tumours (FoundationOne CDx). The test is designed to provide clinically actionable information - both to consider appropriate therapies for patients and to understand the results with respect to resistance - based on the individual genomic profile of each cancer patient. Samples from the tumour are analysed using methods based on so-called *next generation sequencing*. The service is FDA-approved and has national coverage for qualifying Medicare and Medicare Advantage patients across all solid tumours in the US. In Sweden, The New Therapies (NT) Council has recommended that FoundationOne CDx should not be used *routinely* in the Swedish healthcare system, since they found that there are currently no clear advantages over the genetic diagnostics methods that are currently carried out at a lower cost.[4] Also, the genomics medicine centres at the university hospitals seem to be willing to develop that type of service further and to build in-house expertise in the field.[5]

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[3] Personalized medicine at FDA, a progress and outlook report published be the personalized medicine coalition http://www.personalizedmedicinecoalition.org/Userfiles/PMC-Corporate/file/PM_at_FDA_A_Progress_and_Outlook_Report.pdf

[4] Recommendation from the new therapies council in Sweden (FoundationOne CDx, NT-rådets yttrande till regionerna 2019-11-06, in Swedish) https://janusinfo.se/download/18.2d18948e16e3554fc2332f4b/1573021669018/FoundationOne-CDx-191106.pdf

[5] Comments from Genomic Medicine's reference group regarding recommendations for FoundationOne CDx (Kommentarer till "Underlag för beslut i regionerna: FoundationOne CDx, in Swedish)" https://janusinfo.se/download/18.7caa7f2416d85e44a5752f82/1570185523050/Kommentarer%20fr%C3%A5n%20GMS%20expertgrupp%20f%C3%B6r%20solida%20tum%C3%B6rer.pdf

Several pharmaceutical companies are also currently investing in *gene therapy*, which has a specific niche within the precision medicine field. Gene therapy seeks to modify genes in a patient, or introduce new genes, with the goal of treating, preventing or potentially even curing specific diseases, including several types of cancer, viral diseases, and inherited disorders. Gene therapy approaches include replacing a mutated gene that causes disease with a functional copy, or introducing a new, correct copy of a gene into the body in order to fight disease.[6] It could be claimed that gene therapy is one of the most advanced forms of precision medicine.

In the period up to August 2019, 22 gene therapies had been approved by the drug regulatory agencies in various countries and there is a large number of therapies currently being tested clinically.[7] According to the Alliance of Regenerative Medicine there were 352 new gene therapies in clinical trials in 2019 (111 in Phase I, 209 in Phase II and 32 in Phase III).

USE OF DATA ANALYTICS AND ARTIFICIAL INTELLIGENCE TO ACCELERATE PRECISION MEDICINE

The availability of huge amounts of data and powerful tools for data analysis has been crucial for the development of precision medicine. Fuelled by advances in computational power, theoretical understanding, and an ever-increasing amount of data, the last decade has witnessed the widespread application of artificial intelligence (AI) in every major field of society, including medicine and healthcare. Considering the complexity of the biology and chemistry taking place in our bodies and the many factors that affect our health, AI and associated advanced analytical techniques appear to be essential requirements for maximizing the insight from biological data. It is likely that AI will take precision medicine to the next level, increasing the accuracy and prediction of outcomes for patients, and helping us to understand how best to prevent diseases. Some experts claim that the processes involved in precision medicine are dependent on AI or similar techniques to reach their full capacity. Currently there are many AI initiatives within the

[6] Website https://alliancerm.org/technologies/gene-based-medicine/ (accessed April 2020)
[7] Cui-CuiMa et al. Biotechnology Advances Volume 40, 2020, 107502 https://www.sciencedirect.com/science/article/abs/pii/S0734975019302022

various national health systems; several studies have indicated that AI is on par with medical experts in the accuracy of clinical decisions.[8]

WHAT ARE THE LIMITATIONS OF PRECISION MEDICINE?

The applications of precision medicine are currently mainly in the area of rare diseases and cancer. For some cancer types and a few rare diseases, the new processes have already changed outcomes profoundly. However, the number of patients who have benefitted from these advancements is still quite low. Also, the more scientists find out about the genetics of most common diseases, the more complex the interplay of genes affecting the disease mechanisms appears. Some scientists assert that - at least so far - only niche applications have been found for precision medicine, and even though gene therapy is now becoming a reality for a few rare diseases, the effects of precision medicine in general on public health are miniscule while the costs are astronomical.[9] [10] These critics claim that the emphasis on reducing and limiting biomedical explanations to genetic pathways - so-called genetic reductionism - comes at the expense of other important molecular, cellular, physiological and epidemiological approaches.

CAN GENETIC DATA BE USED IN THE PREVENTION OF DISEASE?

As mentioned above, the concept of precision medicine includes the use of genetic and other health data for disease prevention, usually by assessing the risk of developing specific diseases. This aspect of precision medicine is currently very limited in healthcare settings. However, it is used for example in certain types of breast cancer that heavily depend on a specific genetic variation. Individuals that have a family history of that specific cancer type can be genetically tested and may then choose to have surgery to remove their breasts as a preventive step.

Estonia has taken a slightly different approach to preventive precision medicine, currently using genetic screening as a basis for disease prevention, as discussed further below.

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[8] Xiaoxuan Liu MBChB et al., The Lancet Digital Health, Volume 1, Issue 6, October 2019, 271. https://doi.org/10.1016/S2589-7500(19)30123-2

[9] Michael J. Joyner, Nigel Paneth. Precision medicine's rosy predictions haven't come true. We need fewer promises and more debate, STAT News, February 2019 https://www.statnews.com/2019/02/07/precision-medicine-needs-open-debate/

[10] Perspectives in Biology and Medicine, Special Issue on the Precision Medicine Bubble, editors Nigel Paneth, Michael J. Joyner Volume 61, Number 4, Autumn 2018 https://muse.jhu.edu/issue/39661

Several companies also offer genetic tests direct to consumers, allowing testing for gene variations that have been implicated in a higher risk of developing various diseases, including several forms of cancer, diabetes and cardiovascular disease. The validity and usefulness of these direct-to-consumer tests have been widely criticised by scientists. The genetic profiles of most diseases are often very complex, with variations in hundreds of genes which then interplay in development of the disease – the risk factors are polygenic (see below). Critics suggest that it is not possible to draw conclusions from a test that involves only a small number of gene variations, which is what most companies offer. The test result could give the user a false sense of security if only a limited set of "risk genes" is tested for.[11] There are also studies showing that the so-called SNP-chips frequently used in this type of direct-to-consumer genetic testing often give false-positive results for rare pathogenic variants, i.e. wrongly stating that there is a higher risk.[12]

However, the large amounts of data that have been collected by some of the direct-to-consumer gene-testing companies have been used in research that has led to the discovery of potential new drug targets and drugs. In January 2020, it was announced that 23andMe Inc., a company known for its ancestry DNA tests, has licensed an antibody it developed to treat inflammatory diseases to the Spanish pharma company Almirall SA. Before the contract, 23andMe identified the drug candidate and conducted animal studies on that drug. This is the first time that the company has directly sold a product it created using the genetic information collected from their customers. However, earlier, 23andMe had shared genetic data with pharmaceutical companies. In 2018, GlaxoSmithKline signed a contract with 23andMe to use data from customers for drug development and purchased a $300 million stake in the company.[13]

[11] Website, in Swedish https://www.forskning.se/2018/12/17/gentester-lovar-runt-och-haller-tunt/ (accessed April 2020)
[12] Article in the Guardian published 21 July 2019 https://www.theguardian.com/science/2019/jul/21/senior-doctors-call-for-crackdown-on-home-genetic-testing-kits, Michael N Weedon et al., bioRxiv, posted November 11, 2019 https://www.biorxiv.org/content/10.1101/696799v1
[13] Press-release 23andMe https://mediacenter.23andme.com/press-releases/23andme-signs-a-strategic-agreement-with-almirall/

WHAT IS MEANT BY GENETIC RISK?

The contribution of a person's genetic setup to their risk of developing certain illnesses or diseases can be described as their genetic risk. Most common diseases have a genetic component; this means that variations in specific genes can increase the risk of developing the disease. Enhanced screening programmes and focused preventive therapies for a given disease can be achieved by understanding these genetic variations and identifying individuals at high risk of the disease. For a few identified diseases rare so-called monogenic traits, where there is a change in only one or a few genes, can increase the risk of getting the disease several-fold. However, most diseases with a genetic component are polygenic in nature and depend on variations occurring in many genes that have smaller effects and act over long periods of time, often in concert with environmental factors. The cumulative risk, derived from aggregating the contributions of the many DNA variants associated with these complex diseases, is referred to as the polygenic risk score. Studies have shown that, for some diseases, a significant proportion of the population carries polygenic variations that increase the risk of developing the disease threefold. One study showed that eight percent of the studied population had polygenic variations that gave a threefold increased risk of developing coronary artery disease, which was 20-fold higher than the carrier frequency for rare monogenic mutations that conferred a comparable risk.[14] Since the direct-to-consumer genetic tests that claim to identify an increased risk of developing several common diseases often only check for variations in one or a few of the genes known to affect the risk, the value of such tests can be questioned.

It is important to keep in mind that the actual development of a disease most often also depends on environmental factors. For example, data from the Estonian Biobank have shown that the incidence of diabetes in individuals with a large number of genetic variations linked to type 2 diabetes is not higher if their body mass index (BMI) is normal (<25). However, the risk of type 2 diabetes is higher in these individuals if their BMI is higher, and the occurrence of type 2 diabetes increased with an increased number of genetic variants linked to the disease.[15]

[14] Schork, A.J.et al., Genetic risks and clinical rewards. Nat Genet 50, 1210–1211 (2018). https://doi.org/10.1038/s41588-018-0213-x
[15] Läll, K., et al., Personalized risk prediction for type 2 diabetes: the potential of genetic risk scores. Genet Med 19, 322–329 (2017). https://doi.org/10.1038/gim.2016.103

WHAT IS THE STATUS OF PRECISION MEDICINE INTRODUCTION IN SWEDEN AND OTHER COUNTRIES?

Since president Barak Obama launched the Precision Medicine initiative in 2015, backed by the US government with 215 million USD, investment in precision medicine has increased worldwide. The EU has invested heavily through the framework programmes and, in addition to European collaboration projects, most European countries have national initiatives.

Sweden was early in adopting the use of genome sequencing in healthcare to diagnose rare diseases and cancer. In cancer care, about 10,000 genetic analyses are performed every year to help optimise treatment for the patients. The goal now is to boost this capacity to 65,000 genetic samples per year from patients with cancer or rare diseases. Genomic Medicine Sweden (GMS) was launched in 2017 in order to enable the shift to precision medicine nationally and to increase access to testing. GMS a national infrastructure whose goal is to implement precision medicine in clinical settings and strengthen the collaboration between Swedish healthcare, research and the life-science industry. GMS originated in the Clinical Genomics facilities at SciLifeLab, which work to develop and validate new sequencing methods that are highly significant to precision medicine. Important is to develop a national genomics platform, which facilitate the use of genomic data in healthcare, research and for innovation. GMS covers seven regions with university hospitals and universities that have medical faculties. Together with patient organizations, healthcare providers, universities and the industry, GMS aims to transform genomic innovation, based primarily on advanced next-generation sequencing technologies, to enable greater precision in the diagnosis, treatment and management of patients with rare diseases, cancer or infectious diseases.

As in Sweden, most countries have focused their precision medicine investments on cancer and rare diseases. However, some countries (such as the UK and the US) are planning also to invest in precision medicine for prevention, with the aim of starting to offer genetic analysis to healthy individuals. Estonia, a forerunner in this field, has taken a different approach to their national precision medicine initiative compared to other countries. The Estonian population is offered free DNA tests in order to identify people at risk of developing certain diseases. The aim of the initiative, developed in a collaboration

between the Ministry of Social Affairs, the National Institute for Health Development and the Estonian Genome Centre at the University of Tartu (which hosts the Estonian Biobank), is to give lifestyle advice to participants who have a genetic risk of developing conditions such as diabetes, cancer and cardiovascular disease, to prevent future illness.

The decision to start the initiative was taken in 2018 when the scientists at the Estonian Biobank concluded that there was enough knowledge about genetic risk factors to start to give advice to citizens based on genetic tests. The tests have been designed to determine 700,000 genetic variants with the aim of:
1) preventing common diseases (cardiovascular disease and type II diabetes) and breast cancer;
2) searching for known pharmacogenetic mutations (genes known to influence drug response);
3) screening for mutations that increase the risk of rare diseases.

The tests target rare *monogenic variations* and provide risk scores for common diseases based on *polygenic variations*. The budget from the government corresponds to 50 Euro per tested person, meaning that the programme has to be very cost effective. To date, the cohort size is close to 200,000 participants (≥18 years of age), which corresponds to 15 % of the Estonian population. The cohort closely reflects the age, sex and geographical distribution of the population.[16] The participants sign a broad informed consent form that enables the biobank data to be used for research. The consent form also allows researchers to follow the participants electronically on a long-term basis. This means that it is possible for researchers to investigate correlations between genetic data and data from registers containing healthcare visits and prescribed medications. Access to the database for research purposes is clearly regulated in the Human Genes Research Act.[17] The objectives of this act are to "regulate the establishment and maintenance of the Gene Bank, to organise the genetic research necessary therefore, to ensure the voluntary nature of gene donation and the confidentiality of the identity of gene donors, and to protect persons from misuse of genetic data and from discrimination based on interpretation of the structure of their DNA and the genetic risks arising therefrom". Hence, the act sets a framework for the use of genetic data for research, contributing to the establishment of trust in the testing and handling of data among the participants.

[16] Website https://genomics.ut.ee/en/about-us/estonian-genome-centre (accessed April 2020)
[17] Human Genes Research Act https://www.riigiteataja.ee/en/eli/531102013003/consolide (accessed Oct 2019)

After the genetic tests are analysed, individuals with mutations that indicate a higher risk of breast cancer or familial hypercholesterolaemia (FH) - which results in high cholesterol levels and an increased risk of cardiovascular disease - are called to another appointment, to have a new test to confirm the results. After that, the person has a face-to-face meeting with medical experts, a clinical examination and so-called "genetic counselling" where the results and the meaning of the results are discussed. Participants also receive information that can be shared with their families to promote genetic testing of family members.

Results from the Estonian initiative have shown that the mutation carriers have often been unaware of the familial background of the diseases; in fact, in the case of FH, adherence to treatment among those who had been diagnosed earlier was low. However, after the genetic screening and counselling, adherence to treatment increased significantly. Estonian cardiologists and oncologists have generally appreciated the value of the genetics-first approach and now promote further development of genetic screening in the country.

After the tests and consultations, the participants have provided positive feedback. The concern that the participants would predominantly be worried if genetic risks were discovered has been proved wrong; instead, the positive effects of knowledge and the possibility of influencing one's own outcomes have been reported among the participants.

CHALLENGES OF USING HEALTH DATA FOR RESEARCH AND DEVELOPMENT OF PRECISION MEDICINE IN SWEDEN

With possibilities comes challenges; with respect to the use of health data for precision medicine, the challenges are primarily related to the data handling, and include concerns about legality, privacy and ethics, as well as economics.

Sweden was early in initiating large-scale studies similar to those in Estonia. By as early as 2009, six Swedish universities had started the collaborative biobank project Life Gene, with the aim of providing a unique longitudinal study on how our genetic makeup, our environment and our life-style choices affect our health. The approach was to collect samples from a cohort of 300,000 Swedes. However, due a rather restrictive

interpretation of national data protection regulations, the initiative was not given the chance to fulfil the expectation of creating a goldmine for research and providing insights into public health, disease prevention and the development of new treatments. In 2011, the Life Gene project was stopped by the Swedish Data Protection Authority. This was because the broad informed consent form signed by the participants was found to be insufficient since they had not been informed which specific research projects their data and samples were going to be used for in the future. Nonetheless, the government considered Life Gene to be of important national interest and instituted the temporary "Life Gene law", allowing the project to continue.[18] The period of validity of the law has since been extended and it is currently in effect until the end of 2020; however, the scope and impact of the project has been limited because of the legal issues.

In 2012, the Swedish government decided that all citizens should be offered a health account (*Hälsa för mig)* and included the cost for the required infrastructure in the national budget. The investment was intended to give Sweden's residents a free personal health account for lifelong saving, management and sharing of their health data, originating from healthcare institutions and elsewhere. It was also intended to be a platform on which companies and organizations could build innovative e-health services. The aim of the initiative was to strengthen the individual's participation in their own health situation and to give the individual the right and opportunity to dispose of their own health data. An infrastructure solution was tendered in 2013 but, in 2016, the Data Protection Authority claimed that the solution was not compliant with the Swedish legislation on handling of personal information. The initiative was abandoned in 2018 as a result of these legal challenges.[19]

The development of secure, effective handling of genomic data in Swedish healthcare is now a priority in GMS. The goal is to build an infrastructure that can store and organize genetic data, which is also combined with other types of health data. Effective ways of sharing data between caregivers and with researchers, whilst protecting personal integrity, need to be in place. Several countries, including Finland and Denmark, have modified their legislation regarding health data to improve correlations with modern technologies and digital information handling as used today. Their updated legislation

[18] Swedish Law. Lag (2013:794) om vissa register för forskning om vad arv och miljö betyder för människors hälsa.
[19] Website https://www.ehalsomyndigheten.se/tjanster/ (accessed April 2020)

enables the use of data collected in healthcare for research and development of new therapies as well as other types of innovation. Several stakeholders, including caregivers, healthcare professionals and researchers at universities and pharmaceutical companies, have pointed out that the Swedish legislation is difficult to interpret and that it hinders the benefits of sharing data with the aim of improving healthcare and accelerating the research and development of new therapies.[20]

The Swedish national life science strategy presented by the government in December 2019 also points out that better utilization of healthcare data is a prerequisite for Sweden to be able to take the lead in life science. The strategy highlights that the use of health data for research and innovation must increase and that effective and secure exchange of health data is necessary. The Swedish legislation regarding the storage of information in biobanks and the exchange of patient data between caregivers is currently the subject of government enquiries, and a statement is expected to be presented in early 2021.

In Stockholm, during the fall of 2019, the Region Stockholm Assembly, which is responsible for healthcare in the Stockholm region, started to develop a Center for Health Data, with the aim of utilizing health data to enable precision prevention and treatment as well as novel research. The Swedish Data Protection Authority, at an early stage, pointed out the risks associated with this initiative in relation to data security.

National biobanks and health data registers are often highlighted as a Swedish asset for medical research. However, the accessibility and therefore also the usability is also pointed out as being challenging. With other countries now modernizing their legislation at a rapid pace to increase access for research and development, Sweden is lagging behind in this field.

THE PATH FORWARD

If Sweden aims to use healthcare data and data from genetic screening for precision medicine and other research, the legal framework needs to be revised and open public discussion is required on how to judge the health benefits for individuals and society

[20] Website https://medicinskaccess.se/artiklar/precisionsmedicin-nya-moejligheter-att-staella-diagnos-och-faa-raett-behandling/ (accessed April 2020)

against the risk of misuse of gene data in that context. There are certainly risks related to the more open sharing of vulnerable data. What happens if your health data get into the wrong hands and are used to disclose your genetic predisposition to a certain disease or condition to, say, an insurance company or your employer? What happens if your integrity is offended for a purpose that does not benefit you? Who *is* the right person to know what is considered beneficial to you – yourself, a healthcare professional, or a government official from the data protection agency?

On the other hand, what are the consequences of *not* using the available data and tools for improving healthcare, of not developing new treatments and of not working for prevention? With medical breakthroughs in precision medicine, with the research frontier moving rapidly towards in-depth knowledge of a palette of diseases and conditions, with the strive towards equal healthcare – is it ethical not to use the potential of health data? A balanced judgement of value against risk – as always when new strategies are implemented – is called for.

There is a strong need for open and constructive discussions on balancing the benefits of utilizing health data in a more effective way in healthcare, science and innovation with the risks of misuse of the data. Also, the public needs to be included and to have their say. In a public survey carried out by the organization *Forska! Sverige*, 95 % of respondents were positive about sharing their health data for research and purposes aiming to promote health.[21] To investigate public opinion in more detail, WellcomeTrust has initiated a global online survey which will gather public attitudes about genomic data sharing.[22]

The theme for the "The future of Life Science" symposium in 2021 is *The data revolution in life science and healthcare.* The aim is that the symposium's discussion topics will address several of the questions raised in this report.

[21] Report from Research!Sweden (2018): Healthcare Data – High Time for Action
http://www.forskasverige.se/wp-content/uploads/SR-eng-agendarapport-varddata-2018.pdf
[22] Webpage https://societyandethicsresearch.wellcomegenomecampus.org/project/your-dna-your-say(accessed April 2020)