BETTER, SMARTER, NOW:

Personalised Health

Results of the Academy Programme Personalised Health – From Genes to Society (pHealth)

PREFACE

Personalised Health puts the individual in focus. Personal health care approaches are tailored based on specific information on your genes, your body and your lifestyle, to be as effective as possible and to comprehend disease treatment, prevention, diagnostics and rehabilitation. Recent developments in molecular sciences and imagining have opened new avenues for characterising individuals in detail, for a better understanding of the aetiology of diseases and the role that the environment plays. Major advances in information technology make it possible to analyse, share and store this growing amount of information. Given that the regulatory environment is encouraging, this information profoundly changes the health care system. The introduction of tailored treatments results in improved health while minimising both suffering and the spending of time and expenses.

Finland, and the Nordic countries in general, have several strengths to facilitate the realisation of personalised health. Among our assets are high-quality biobanks, health care registers and personal identification numbers that make it possible to carry out longitudinal research based on a combination of data from different data sources. In addition, we have a top-quality health care system, a population that is generally positive towards research, and excellent research environments in the health research area.

On these premises, the Academy of Finland launched the Academy Programme *Personalised Health – From Genes to Society* (pHealth) in 2015 and provided 11 million euros in funding for nine scientifically excellent research projects in the open programme call. The first and foremost theme of the research programme is to produce

Personalised Medicine "refers to a medical model using characterisation of individuals' phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention".

According to: Horizon 2020 and European Council Conclusions on peronalised medicine for patients (2015/C 421/03)

data and tools to contribute to the understanding of individual characteristics at the molecular level, and to use these for health promotion. The objective is to implement the programme in the best interests of basic research and to help individuals and society benefit from using genetic information and genetic health data. The funded projects represent genuinely multidisciplinary consortia that bring together different scientific disciplines with a view to understanding new kinds of research perspectives such as studies that move from basic research to research targeting individuals, the healthcare system, business companies or society at large.

Since the programme was launched, the interest towards Personalised Health has grown both in Finland and internationally. As Personalised Health concerns both the health care system and the health industry

and business sector, the Academy actively collaborates with many stakeholders. The most important forum for national collaboration is the *National Health Sector Growth Strategy for Research and Innovation activities*. International collaboration is advanced both at the Nordic (NOS-M and Nord-Forsk) and the European level (ICPerMed).

This publication was released in November 2019 for the seminar **Better, Smarter, Now: Personalised Health**, where the Academy presented the results of the pHealth programme and its nine research projects. We hope this publication will give you a view on the impact that excellent research can have on your personal health as well as on our society and the health care system in a broad perspective.

Personalised health is not a distant utopia: it's already here. Why is personalised health an essential area of research and innovation? Three experts of personalised health share their views on the topic.

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At the moment there are certain medical problems that will become more common, such as obesity, diabetes, and various cancers. It is crucial to invest in research of the diseases that are an increasing burden so that we can treat them more accurately and effectively. As these diseases are challenging us globally, we in Finland have a great opportunity to help create new, more personalised treatments. Our high-quality research on personalised health could and should be put into use all around the world.

However, the way forward is far from uncomplicated. In a world of fake news and a constant flow of information, we need to strive hard to really introduce new knowledge into people's everyday life. Despite these challenges, I see a bright future for personalised medicine."

We are in a situation where opportunities to develop personalised health are more abundant than ever before. The new knowledge we gain about an individual's genome, its function, cellular biochemistry and physiology changes the whole paradigm of healthcare. Since the information of an individual's genome touches upon one's relatives and is highly personal, we need to consider very carefully how we use this information. This is where we need legislation, in other words, the state comes in.

The Ministry of Social Affairs and Health, in cooperation with national and international experts, is actively preparing practices and regulations for using genome information. Finland has already enacted a law about the secondary use of health and social data, but we are constantly developing these matters and setting common goals, which can also be discussed at EU level. We want to be forerunners in solving the ethical and practical issues considering data privacy and protection."

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❝❝ At the moment, oncology and rare diseases drive personalised medicine. This is because these are areas where targets for intervention and diagnostic tools are either already in place or novel biomarkers are constantly being identified through modern genetics. The challenge is to transfer technology and approaches to other non-communicable diseases and multifactorial disorders including complex patterns of diseases, of which many affect the elderly.

The Nordic countries have a number of advantages to facilitate the realisation of personalised medicine and health, for example high-quality epidemiology and clinical research, as well as unique health data infrastructures in registers, cohorts and biobanks. We have similar public health care systems and a population that is generally very positive towards participating in research. People also have trust in research which brings a strong added value in collaboration between the Nordic countries."

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Personalised medicine to predict and prevent type 1 diabetes

P4 Diabetes

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New insights into predicting and preventing type 1 diabetes

Type 1 diabetes (T1D) is so far an incurable disease constituting a substantial burden to individuals. T1D poses a great challenge to the health care system in all developed countries due to its high prevalence and serious long-term complications. Better understanding of disease pathogenesis and stratification of heterogeneous disease is central to improving means to predict, diagnose and monitor T1D as well as to developing personalised therapeutic and prevention strategies.

T1D is characterised by the loss of insulin-producing pancreatic beta cells. We discovered that beta-cell function is impaired already several years before diagnosis of clinical T1D. Factors associated with the rate of disease progression were identified. Changes in the composition of immune cells after appearance of autoantibodies to clinical T1D were detected. We confirmed the previous findings indicating that the islet autoimmunity initiated by either IAA or GAD antibodies can be considered as distinct endotypes of T1D.

We found associations with infections, microbiota and diet. Vitamin-D appeared not to influence the disease process.

We confirmed our previous findings indicating that diminished specific phospholipids precede islet autoimmunity in children who later develop clinical T1D. Potential explanation for this may be prenatal exposure to per- and polyfluoroalkyl substances (PFAS), which was found to associate with increased T1D risk.

We established a reference model for the evolution of a healthy serum proteome during early childhood and found age to influence approximately half of the studied proteins. Changes associated with the development of T1D were identified for further validation. We discovered a gene signature detected early, already before the appearance of T1D associated autoantibodies, that could help in identifying those children who are likely to develop the disease later.

Legal and ethical challenges related to personalised medicine were studied and a paper

- Type 1 diabetes (T1D) is the most common endocrine disease among children. It is a chronic immune-mediated disease characterised by selective loss of insulin-producing pancreatic beta cells in genetically predisposed subjects.

- Less than 10% of genetically predisposed individuals progress to T1D emphasizing the importance of environmental factors in the disease process.

- The appearance of T1D-associated autoantibodies is a first sign of T1D, but it takes from a couple of weeks to decades to the clinical presentation of T1D. Lifelong insulin is the current therapy.

on this topic, edited by Raimo Lahti and Juli Mansnérus, is in preparation.

Longitudinal carefully controlled characterisation of children developing T1D revealed heterogeneity and distinct endotypes based on autoantibody profiles. Early changes in immune cells in children who later developed T1D were detected already before the appearance of T1D associated autoantibodies. A great amount of evidence supports the importance of early life. Further studies will focus on longitudinal deep molecular profiling of children with carefully defined phenotype during the first years of life. Validation and integration of the key findings will provide basis for stratification of children for new therapies and prevention.

**Personalised combination therapies
for high-grade serous ovarian cancer patients**

OVCURE

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Computational tools improve
the treatment of ovarian cancer

Ovarian cancer kills over 184,000 women yearly. The standard treatment for the most common subtype, high-grade serous ovarian cancer (HGSOC), is surgery and platinum-taxane combination chemotherapy. While most patients initially respond well to the treatment, it is seldom curative, and more than 50 per cent of the patients die within five years.

The main question in oncology is whether a patient will respond to the first-line therapy or not. A way to reliably predict therapy response allows closer follow-up and changes to inefficient therapy. To enable this, we developed a cloud-based machine learning system (CLOBNET). CLOBNET uses patient's electronic health records and clinical data, and is already able to predict poor or good response with 86 percent accuracy. We can detect women at risk of developing drug resistance at early treatment stages without waiting for biomarker and imaging results, and months or years before clinical symptoms relapse.
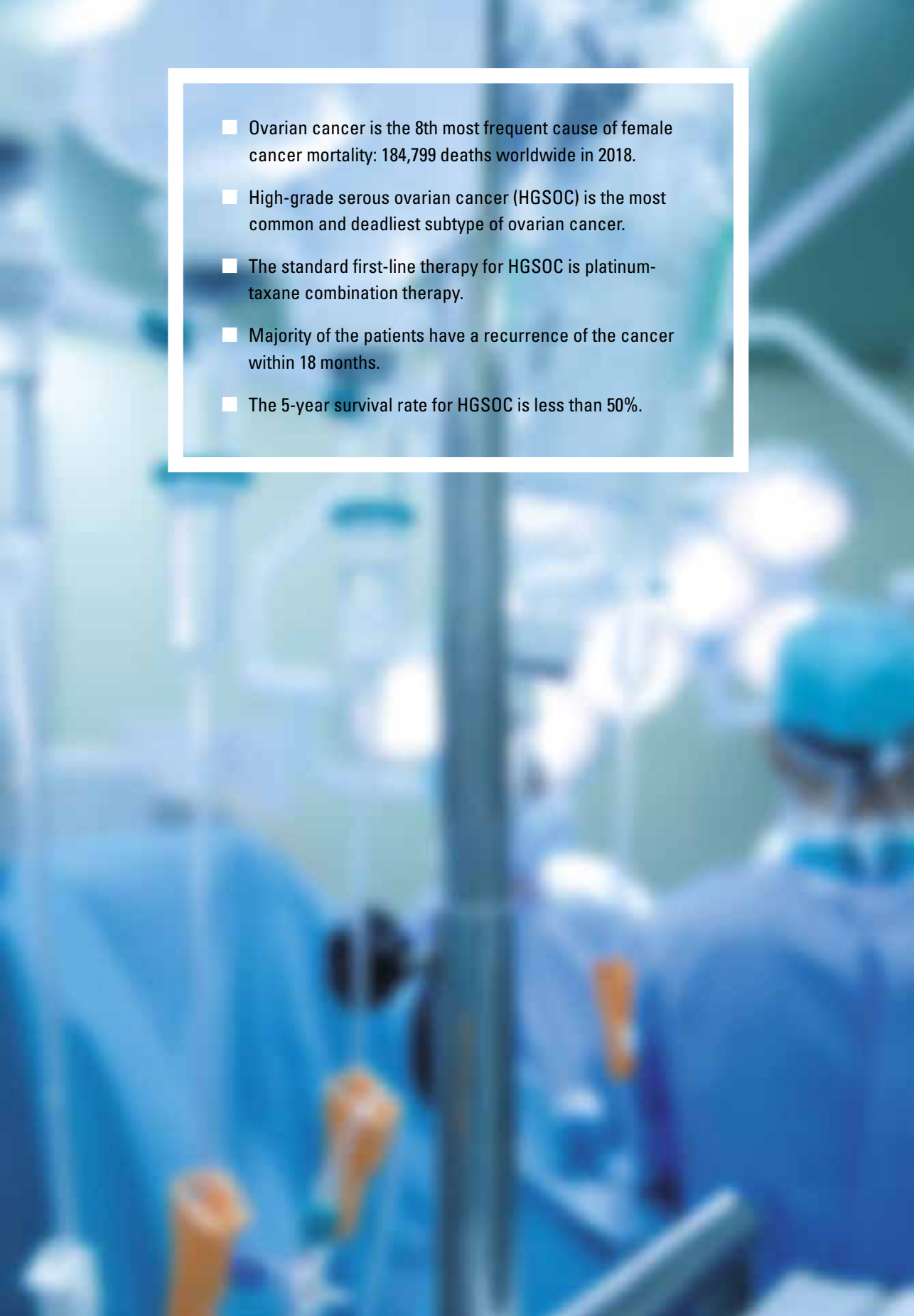
Another key issue is to identify resistance mechanisms and design interventions to overcome them. We developed a mathematical model for tumor evolution, based on time-series data from HGSOC patients, which allowed for the first time to reliably estimate the tumor-cell killing effect of platinum in patients. Our results show that while at diagnosis there are on average five active resistance mechanisms, they are not equally distributed; typically only one or two mechanisms dominate. This is an important observation and facilitates the planning of effective combination therapies.

To find suitable therapies, we developed several computational tools that can be used to test and predict drug combination effects in patient-derived cells. In such a highly heterogeneous cancer, validation of the computational results is crucial; to this end we have collected and imaged over a hundred pathology slides from HGSOC tumors.

The methods developed in this project can lead to improved patient survival when applied in the clinic. They can also assist the pharmaceutical industry and clinical researchers to design clinical trials and select drugs that show improved efficacy in patient groups classified according to molecular subtypes.

- Ovarian cancer is the 8th most frequent cause of female cancer mortality: 184,799 deaths worldwide in 2018.

- High-grade serous ovarian cancer (HGSOC) is the most common and deadliest subtype of ovarian cancer.

- The standard first-line therapy for HGSOC is platinum-taxane combination therapy.

- Majority of the patients have a recurrence of the cancer within 18 months.

- The 5-year survival rate for HGSOC is less than 50%.

Exome sequencing is effective in diagnostics of neurological diseases

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Clinical exome sequencing is effective as a first-line diagnostic tool in neurological diseases

Finding a diagnosis for a patient with neurological symptoms is often complicated and expensive, and typically requires extensive investigations by specialised doctors. Reaching an exact diagnosis is important as it puts an end to numerous examinations and diagnostic tests, settles the cause of disease and its natural course and gives it a name. In some rare cases, knowledge of the molecular cause of a disease opens possibilities for personalised therapies.

Many neurological diseases belong to rare disorders, and are caused by gene mutations. In our project, we studied the effectiveness of clinical exome sequencing as a first-line diagnostic tool in neurological diseases. Clinical exome sequencing allows the investigation of all known disease genes in a patient's genome at once.

We studied how well exome sequencing works when a new patient with a potentially genetic disease enters the neurology clinic. We investigated how likely the exome would identify the disease cause and reduce the need for other diagnostic investigations.

If the exome were the first diagnostic test, would it be cost-effective? How would it impact patients' lives?

We performed clinical exome sequencing on 50 recruited pediatric patients and 100 adult patients. We built a bioinformatics analysis platform for analysing the clinical exomes for neurological diseases. Confirmed diagnosis was reached for nearly 30 per cent of the patients, with a higher probability of findings for children in comparison to adult patients. Within these we found for example a novel cause for an adult-onset muscle disease and mutations underlying PURA syndrome in two children.

We also compared the costs of the diagnostic routine between the participating patient group and a retrospective patient group. We conclude that clinical exome sequencing is cost-effective at its current price as a first-line diagnostic tool in neurological diseases. However, cost-effectiveness is sensitive to prices of operations, the patient group and age.

- The complexity of the human brain and nervous system gives rise to more than 600 different neurological disorders.

- The manifestations vary from early-onset brain disorders of childhood with epilepsy, progressive encephalopathy and early death, to progressive but milder adult-onset degenerative disorders of peripheral nerve, muscle and brain.

- Overall, these diseases cause immeasurable suffering to individuals and families, and a vast economic burden to societies in medical and social expenses and lost productivity.

- The total cost of brain disorders in Europe is estimated to be 800 billion euros per year.

Collecting knowledge of the gene mutations underlying neurological diseases in Finland is important for the implementation of personalised genome testing into the health care system. Based on this study, recommendations on the use of clinical exome sequencing can be given for different patient groups. Scientific reporting of the identified mutations, the cost-effectiveness analyses and the results of questionnaires that we used for studying the patients' and their guardians' attitudes towards genetic testing are still ongoing.

**Finnish cardiomyopathies –
from genetics and phenotype to pathogenesis**

CardioPath Study

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Unique mutations cause hypertrophic cardiomyopathy in Finland

Cardiomyopathies are major causes of mortality and morbidity, especially for young people. Our study investigates Finnish cardiomyopathies and their genetic background, clinical phenotype, prognosis, and molecular and functional changes in induced pluripotent stem (iPS) myocyte models.

The genetic background and pathogenesis of cardiomyopathies are currently a target of keen interest, as modern next generation sequencing techniques allow rapid analysis of disease-causing mutations. iPS myocyte models, in turn, enable the research of pathogenesis of cardiomyopathies at a new level.

We have identified the genetic basis and outcome of hypertrophic cardiomyopathy (HCM), the most common hereditary heart disease in Finland. By targeted next-generation sequencing techniques, we identified disease-causing mutations in 38 per cent of 382 Finnish patients with HCM. Uniquely, four major sarcomere mutations accounted for 28 per cent of HCM cases, whereas HCM-related mutations in non-sarcomeric genes were rare. Mortality in patients with HCM was not high but exceeded that of the general population. In Finland, there are at least 27 HCM-related deaths annually.

We have also established an iPS cell library for the major mutations causing HCM in Finland, and a databank combining clinical and genetic data of the patients with the mutation-specific iPS cell lines. We have established a mathematical platform to improve the translation from hiPSC-CMs to humans.

In mutation-specific phenotype characterisation of Finnish cardiomyopathies, we have identified several ECG, imaging and metabolomic features, which differentiate between mutation carriers and healthy individuals. On the other hand, these features characterise severe phenotype with arrhythmia vulnerability. Our studies have had a positive impact on diagnostics and clinical treatment of patients with cardiomyopathies in Finland, and we have educated professionals and the public on this major health problem.

At present, we investigate the genetic basis of dilated, Fabry and transthyretin cardiomyopathies in Finland with next-generation sequencing techniques. We continue to collect an iPS cell library of Finnish cardiomyopathies and a databank combining clinical and genetic data. All this research is crucial for finding the best medical treatment for each mutation.

- Hypertrophic cardiomyopathy is the most common hereditary heart disease and the most common cause of sudden cardiac death in young individuals and athletes.

- Targeted next-generation sequencing techniques and exome sequencing have emerged as an efficient tool to identify new mutations and genes responsible for monogenic diseases.

- Pathogenesis of cardiomyopathies is largely unresolved. The mechanisms by which specific mutations cause cardiomyopathy are poorly understood, and specific treatments for patients are lacking.

- Induced pluripotent stem (iPS) cells can be generated from somatic cells of the patients by epigenetic programming, and differentiated into cardiac myocytes. The iPS cell-derived cardiomyocytes can be used in drug discovery, to study mechanisms of cardiomyopathies and to design patient-specific therapies.

Good(s) for health: Personalized health services and flexible appropriation of bioinformation

GoHe

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The advancement of innovation in personalised health care is predominantly concerned with economic expectations and commercialisation

The work of the research consortium is focused on the most topical issue in personalised health. The main question is: How do the practices of collection, circulation and management of personal health-related data affect the potential and the prospects of personalised medicine in Finland?

We study the utilisation and management of digital samples, patient and population data in biomedical research, clinical practices and business activities advancing personalisation of medicine. Our studies target four settings:

1) biobanks,

2) an experiment to use genomic risk information in clinical practice,

3) collaboration of academic and commercial partners in biomedical basic research and

4) commercialisation efforts related to data collection and management.

The analyses focus on expectations of utilisation of digital health data, flexibility of data, and complexity of data management.

Providing personalised healthcare is as a complex social and political process. It involves people, institutions and multiple practices aiming simultaneously at advancing medical science, implementing new technologies in clinical practices and medical organisations, building innovation and business 'ecosystems' and mobilising expectations about a medical future.

In the Finnish context, the main findings are firstly the promotion of personalised health care takes predominantly place in the framework of innovation policy and is mainly concerned with economic expectations and commercialisation. Secondly, collaborative R&D between commercial and academic partners can extend opportunities and resources of basic research in biomedicine, but collaboration requires a suitable funding framework. Finally, while the rhetoric promoting personalised health implies the engaged and willing population, the implementation of data intensive personalised medicine remains often detached from people's lives and experiences.

A social science perspective of GoHe allows analyses of concrete alignments of biomedical research, health care practices, and commercial pursuits in personalised medicine. In particular, GoHe studies bring relevant knowledge about the patients', customers' and citizens' positions and agency in this development. Our studies also show the social, ethical and economic aspects of utilisation of digitalised bio and health data in evolving domains of personalised medicine. Transnational context of these developments – or the 'global health data economy' – need to be emphasised more in further studies and policy-making.

- Utilisation of masses of digital data, or 'Big Data', from biobanks and other health care data repositories, in biomedical research and clinical practices is a key factor in the development of personalised medicine.

- Biobanks, researchers, hospitals and other healthcare organisations, clinical professionals, public authorities, and private companies in medical business produce, manage and utilise health-related personal and population data. The wide range of producers and users of data make personalised healthcare a complex social and political process.

- Understanding the views of patients and the public is crucial for the advancement of personalised health care and utilisation of digital health data.

Dyslexia: Genes, brain functions, interventions

DysGeBra

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Children with severe risk for dyslexia need more support

Our research focuses on dyslexia, which is a major health-related challenge with a genetic background and is related to abnormal brain activity. The research aims to discover new insights into the etiology of dyslexia and to overcome this challenge by collecting information from genetic, brain and behavioral observations of dyslexia.

Three cohorts of first graders (N=285) with varying risk of dyslexia were recruited for the study in 2016–2018. The development of their reading and spelling skills were followed with behavioral assessments until the end of second grade. Of these children, 184 had severe difficulties in the beginning of the study (reading/spelling below 5th percentile). These children participated in game-based reading interventions delivered by GraphoLearn technology (see grapholearn.info). DNA samples were collected from 138 volunteering children (of which 101 belonged to the severe difficulties group), and 30 children participated in an MEG study on incidental phonological learning during the spring of their first school year. Magnetoencephalography (MEG) is a functional neuroimaging technique for mapping brain activity by recording magnetic fields produced by electrical currents occurring naturally in the brain, using very sensitive magnetometers.

Due to the longitudinal nature of the study, data collection was continued until the spring of 2019, and the results have not yet been ready for publication. Initial analyses suggest that a subgroup of children (circa 30%) with severe reading difficulties responds poorly to the currently available support methods (i.e., special education and GraphoLearn). These children could benefit from additional training of phonological skills, for example in the form of structured spelling practice, which we piloted in our study with promising results. The genetic analysis so far has focused on the comparison of subgroups with different ROBO1 haplotypes.

The results will help the design of intervention methods for children at risk for dyslexia. The new knowledge can be delivered to the special education teachers by training programs, which will guide them for the optimal use of the methods.

- Dyslexia is an unexpected difficulty in learning to read despite normal intelligence, normal peripheral senses and adequate opportunity.

- Dyslexia is the most common learning disability worldwide, affecting between 5–10% of school-age children.

- Epidemiological studies have confirmed a strong genetic influence. Neuroimaging studies have supported the theory that dyslexia has specific, common biological attributes that are independent of the language spoken.

- The Jyväskylä Longitudinal Study of Dyslexia (JLD) showed that about half of the children born at familial risk for dyslexia ends up facing dyslexia. As the JLD's publications have shown, the halves can be identified already at the age of 3–5 days using brain measurements.

Dietary Gluten-induced Disease Entities – from Genomics to Personalized Health and Well-being

DIG IN Health

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Half of celiac disease patients can be diagnosed non-invasively

The methods of personalised health could change the ways of diagnosing and treating celiac disease.

For a long time, the diagnosis of celiac disease has been based on the demonstration of small bowel mucosal damage in biopsies taken upon invasive endoscopy. In practice, this invasive diagnostic practice has followed a "one fits all" type of scheme.

The clinical picture of celiac disease varies considerably from gastrointestinal to extraintestinal symptoms. Also the varying severity of the symptoms makes the diagnosis challenging. Life-long strict gluten-free diet (GFD) is currently the only treatment. Although in most cases the diet is effective, some patients remain non-responsive and some suffer from persistent symptoms despite the diet. Factors contributing to the variation of the clinical phenotype and the response to GFD are obscure.

The aim of the current project was to identify genetic, environmental or downstream biomedical markers that predict the development of celiac disease at the individual level. We also wanted to find out whether these markers can be exploited in predicting the clinical presentation and the response to GFD.

According to our results, the diagnosis of celiac disease can be made in half of the cases without the small bowel mucosal biopsies. This is possible if the circulating serum celiac disease-specific antibodies are high in distinct measurements.

Our results also show that in addition to the certain HLA-type, other genetic variants and an immunological response with distinct characteristics might contribute to the presentation of the disease. The results also show that the response to GFD is affected by the severity of mucosal damage, celiac-specific antibody levels and presence of malabsorption and anemia at the time of diagnosis along with the diagnostic delay. The persistence of symptoms was found to be related with the length of GFD and the fiber content of the diet.

We have already been able to bring some of our results to clinical practice. In December 2018, the current Finnish care guidelines for celiac disease were updated to allow antibody based diagnostics in certain cases. When the diagnosis can be made non-invasively and faster than before, it benefits both the patients and the health care system, saving considerable amount of health care resources. Moreover, the diagnosis and treatment of celiac disease will be facilitated based on the factors affecting the clinical phenotype and the response to the gluten-free diet.

- Celiac disease is an immune mediated condition in which ingestion of wheat, rye and barley induces inflammation and damage to the small intestine.

- The harmful component in these cereals is called gluten.

- Celiac disease is estimated to affect 1 in 100 people worldwide. In Finland, the prevalence is approximately 2% among adults.

- The only effective treatment for celiac disease is a strict life-long gluten-free diet in which all food based on or containing wheat, rye and/or barley are excluded.

Genetic risk information may enhance the effect of health advice

ApoE4mot

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Lifestyle advice increases motivation to improve health behaviour, whereas information about a risk genotype does not have the same effect

Cardiovascular diseases (CVD) and Alzheimer's disease (AD) are common in Finland and will still increase in prevalence as the population ages. Both diseases are affected by lifestyle. Efficient interventions may significantly reduce the high healthcare costs of CVD and AD.

We studied whether information about increased personal genetic risk (here APOE ε4 genotype) to CVD and AD together with dietary and lifestyle guidance motivates study participants from South Ostrobothnia, Finland, to change their lifestyle and whether it affects their health and taste attitudes in a 1.5-year intervention.

We also examined the long-term effects, including potential stress effects, of the disclosure of genetic risk information on health by following up on a group of individuals who received their APOE genotype and health advice in our previous intervention in the years 2010 and 2011.

We also paid attention to ethical issues related to genetic testing. We studied for example the understanding of the given information by using questionnaires.

During the 1.5-year intervention, improvements in the total cholesterol and LDL cholesterol levels and plasma total omega-3 proportions were observed among all participants reducing the risk for cardiovascular diseases and Alzheimer's disease. The APOE ε4 carriers of the intervention group receiving the ε4 carrier status information did not seem to be more responsive to the monthly dietary and lifestyle counselling compared with APOE ε4 non-carriers in the intervention group and control groups.

However, receiving information on increased personal genetic risk provided long-term motivation for improvements in health behavior. The resulting changes, while modest, remained visible even after a number of years.

- Cardiovascular diseases and dementias, including Alzheimer's disease are two of the top three most common causes of death for adults in Finland.

- Risk of cardiovascular and Alzheimer's diseases can be reduced significantly by adhering to recommendations for healthy lifestyle.

- Apolipoprotein E (APOE) ε4 allele is associated with higher risk for these diseases compared with ε3 and ε2. Thus the interest in combining personal genetic information and personalized nutrition in disease prevention has increased, but the research in this area is still scarce.

- The worldwide allele frequency for the ε4 risk allele is about 13.7% whereas in Finland the allele frequency is about 30%.

- Ethical issues in connection with nutrigenetic studies have seldom been pursued. The development of questionnaires, such as studied in this project, would be of great help for conducting ethically sustainable research in the area.

According to the ethics questionnaire there were no differences in the responses depending on the educational level of the participants. The participants were satisfied with the amount of given information, the way information was given and the time given for consideration. In the feedback we collected at the end of the study, the participants expressed their satisfaction about the study in general.

This information is vital for improving the efficacy of health education in order to prevent lifestyle-related diseases that are becoming increasingly prevalent in the developed world.

Action on Human Surfaces: Wound-Metagenomics

Bugsy

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Understanding surgical site infections by accurate sequencing-based diagnostics

Hospital acquired infections are a serious public health problem. Surgical site infections (SSIs) are one of the most frequent types, accounting for up to 20–30% of all hospital acquired infections. SSIs occur in approximately 2–5% of surgical interventions and substantially increase treatment cost, length of hospital stays, morbidity, and mortality.

SSIs are caused by a versatile group of pathogens. The mechanisms by which they cause SSI is not well understood, and their management relies mainly on antimicrobial therapies chosen by trial and error.

Our pHealth consortium brought together experts in surgery, wound healing, microbiology, bioinformatics and computational genetics to develop new strategies for a more accurate diagnosis of microbiological infections. We developed a novel metatranscriptomics-based assay for the comprehensive characterisation of microbes and their activities in complex host-microbe samples and established ultra-fast algorithms for the mining of pathogens from gigantic sequencing datasets. We applied these methods to nearly 50 SSI cases and demonstrated the value of modern genetic solutions in understanding SSIs.

The Helsinki University Hospital has adopted our innovations and has applied them to hundreds of additional infection samples. Our multidisciplinary research team also expanded beyond SSIs and found an unmet need for accurate sequencing-based infection diagnostics in cancer research. In the future, we will keep improving our methods further; we strongly believe that they will become an integral tool for the analysis of microbiological pathogens and their antibiotic resistance activities in complex infection cases.

- Surgical site infections constitute up to 20–30% of all hospital-acquired infections.

- In Finland the cost estimate for treating the infections ranges from 63 million to 200 million euros per year.

- In the United States there are 500,000 cases of surgical site infections each year leading to 8,200 deaths. The cost estimate for treating the infections rises up to 1.6 billion dollars per year in the United States.

Health from Science
(TERVA) 2018–2022

The Health from Science (TERVA) Academy Pro gramme focuses on researching and solving health issues related to major public health diseases. There are still a number of unsolved questions concerning a great many diseases that cause a major burden on the quality of life, the society and the economy. The goal of the programme is to seek bold, new research initiatives for treating these diseases. From the viewpoint of public health, sig nificant breakthroughs often emerge from unexpect ed results that are firmly based on high-quality basic research and risk-taking.

This Academy Programme highlights the impor tance of scientific research in solving public health challenges. In addition to science renewal and high impact, the programme also aims at foster ing new kinds of collaborations between funding agencies and foundations that support research into diseases. There are four foundations involved in the programme: the Finnish Brain Foundation, the Foun dation for Pediatric Research, the Finnish Medical Foundation and the Cancer Foundation.
aka.fi/terva

Health from Cohorts and Biobanks
(COHORT) 2017–2020

The Health from Cohorts and Biobanks (COHORT) Academy Programme promotes research collabora tion and integration using Finnish birth cohorts. In the programme, funding is awarded for integration between projects that already have basic research funding in place. The aim is to increase cooperation between different research partners and especially different levels of research, which through scientific regeneration will contribute to enhancing the impact of research in this thematic area.

The programme enhances the impact of research by promoting more efficient and diverse research uses of birth cohorts: the simultaneous use of multiple approaches and methods will produce new, compre hensive and increasingly applicable information that can be used to develop better methods of diagnosis and treatment, to strengthen the health sector busi ness and to promote public health and health-relat ed political decision-making. The programme will support the effectiveness of national biobanks.
aka.fi/cohort

ACADEMY OF FINLAND