BANBURY CENTER REPORTS

Studying the genomic variation that underlies health and disease: The unique contribution of the Nordic health systems

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Organizers:
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Nelson Freimer, University of California, Los Angeles, USA
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Hakon Heimer, Cold Spring Harbor Laboratory, USA
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BACKGROUND

New sequencing technologies will soon make it feasible to determine, for all individuals in a population, the genomic variations that underlie disease risk and health. However not all countries will be equally prepared to take advantage of this technological revolution, either to achieve scientific breakthroughs or to advance the health of their inhabitants. The Nordic countries are in a unique position to exploit genomic-based medicine. Each country has a public single-payer health system, a strong tradition of nationwide epidemiological research; and health and social records that are unmatched worldwide.

Investigators from the Nordic countries already have initiated international collaborative projects to identify genomic variations that contribute to health and disease. An example is the Sequencing Initiative Suomi (SISu), which has brought together scientists from Finland, other European countries, and North America to develop large-scale medical sequencing projects in the Finnish population, and which has the goal of comprehensively cataloging genome-wide genetic variation in at least 100 000 Finns by 2016.

However, it is increasingly clear that datasets much larger than those available within any single Nordic country will be needed to realize the full potential of medical genomics for science and public health; a comprehensive understanding of the relationship between genetic variation, health, and disease. To achieve such an objective will require coordination of projects and data from across the Nordic countries.

The goal of this meeting was to discuss ways to stimulate coordination of medical genomics across Nordic countries, and specifically to suggest how best to engage the health systems of these countries – and the use of their health records in particular – in such cross-national efforts.

It was expected that the meeting would produce three practical outcomes: (1) identification of possible pilot projects; (2) outlining a white paper describing the advantages of a cross-national collaborative approach to using Nordic health records, and setting out the obstacles that would have to be overcome; and (3) establishing a structure (a working group) for advancing the discussion of cross-national collaborations and for identifying sources of funding for such efforts.

DETAIL

Throughout the course of the meeting, participants were to review: Current projects attempting to incorporate information from Nordic health records for investigations of genomic variation in relation to disease; the organization of Nordic health systems (availability of existing health records, their formats and their accessibility; similarities and contrasts with selected non-Nordic countries); existing ideas for increasing the scale and scope of genomic investigations within the Nordic health systems, including prospective studies; ethical and legal issues affecting cross-national studies (considerations of privacy and confidentiality that are specific to each country, legal differences between each country and other European and North American countries, impact of the fact that the Nordic countries include both EU Members and non-members).

The participants engaged in two-and-one-half days of intensive discussion and, at times, spirited debate about the potential and framework for increasing the level of collaboration among Nordic health researchers in order to meet global health challenges. The focus of the meeting was on leveraging the comprehensive demographic and health data available on more than 26 million people to perform genomic, epidemiologic, and other public health research. To maintain focus, the meeting concentrated on two broad health themes, cardiometabolic disease and mental illness, each of which is among the leading contributors to the burden of disease, both in the Nordic countries and globally.

Attendees described the Nordic countries, with more than 1 million people already participating in various cohorts, as an especially valuable test ground for the US Precision Medicine Initiative, since these infrastructures have already been utilized extensively by teams of US and Nordic researchers for many years. However, these efforts have been piecemeal and often in isolation from one another.

Specific advantages of working with the Nordic health systems include a great deal of public support and governmental support for using the complete demographic, socioeconomic and health registry information for retrospective and prospective research. This has resulted in the initiation of large cohorts that include biological samples. In particular, the Nordic countries have been leaders in creating large biobanks. Finally, the proactive public health systems provide opportunities for innovations based on the knowledge gained.

Some specific projects were mentioned as having a high likelihood of success, with the potential for immediate impact in terms of clinical care or stimulating further high-priority research. These included diabetes, where recent discoveries in genetic variation could lead to preventive strategies and better treatment, and psychiatric research, where recent genomic research has offered some real clues to understanding these burdensome disorders. Another suggested study, one that would take a broader approach, is to catalogue loss-of-function gene variants and their associated phenotypes across the Nordic countries.

Capitalizing on the full potential of the Nordic health infrastructures to study disease will require an ambitious and well organized health research and innovation program that partners with academia, patient groups, governmental institutions, and the private sector. It should certainly build upon existing projects, and although it will be necessary to develop new methods and guidelines for sharing information, these must harmonize with Nordic laws and the ongoing development of Nordic and European Union regulations for data protection.

MEETING SESSIONS

Session 1: Opportunities in Genetics/Genomics
Chair: Ole Andreassen, University of Oslo, Oslo, Norway

Nelson Freimer, University of California, Los Angeles, California, USA
Deep Phenotype Data Obtained Over Decades: Can Genomics Turn Past Investments into Future Health?

Aarno Palotie, Institute for Molecular Medicine, Helsinki, Finland
Special features of Finland for developing genome medicine

Andres Metspalu, University of Tartu, Estonia
Deep sequencing of the Estonian population sample of 2400 subjects - first results

Kristian Hveem, Norwegian University of Science and Technology, Levanger, Norway
Perspective from the Nordic cohorts and registries

Anjené Addington, National Institute of Mental Health, Rockville, Maryland, USA
Perspective from the NIH

Caroline Fox, Merck Research Laboratories, Boston, Massachusetts, USA
Perspective from pharma

Session 2: Opportunites in Data mining and informatics
Chair: Jaakko Kaprio, University of Helsinki, Finland

Søren Brunak, University of Copenhagen, Copenhagen, Denmark
Disease trajectories and time-ordered co-morbidities

Eivind Hovig, Oslo University Hospital, Oslo, Norway
Perspective from the Nordic cohorts and registries

Juni Palmgren, Karolinska Institutet, Stockholm, Sweden
Infrastructure for data - integrating health, lifestyle and molecular information

Jennie Larkin, National Institutes of Health, Bethesda, Maryland, USA
Perspective from the NIH

Morten Sogaard, Pfizer, Inc., New York, New York, USA
Perspective from pharma

Session 3: Opportunities for clinical and translational application of genetics and informatics
Chair: Anu Jalanko, National Institute for Health and Welfare, Helsinki, Finland

Leif Groop, Lund University, Malmö, Sweden
Towards precision medicine in diabetes

Ole Andreassen, University of Oslo, Oslo, Norway
Neuropsychiatric disorders – opportunities for prediction and stratification

Nathan Stitziel, Washington University School of Medicine, St. Louis, Missouri, USA
Identifying and Validating Therapeutic Targets for Cardiovascular Disease

Pål Njølstad, University of Bergen, Norway
Monogenic Disease - Beacons For Identifying Therapy-Relevant Novel Causes of Complex Disorders

Cashell Jaquish, National Heart, Lung and Blood Institute, Bethesda, Maryland, USA
NHLBI Precision medicine/whole genome sequencing program: NHLBI TOPMed

Qingqin Li, Janssen Research & Development, LLC, Titusville, New Jersey, USA
Perspective from pharma

Sessions 4 & 5: Breakout Groups with Presentations/Discussions
Developing proposals for opportunities to collaborate:

Genetics/genomics – Mark Daly, lead
Data mining/informatics – Eivind Hovig, lead
Clinical/translational – Joakim Dillner, lead

MEETING PARTICIPANTS

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