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Pan-cohort metabolomics – The future of population health

Abstract book

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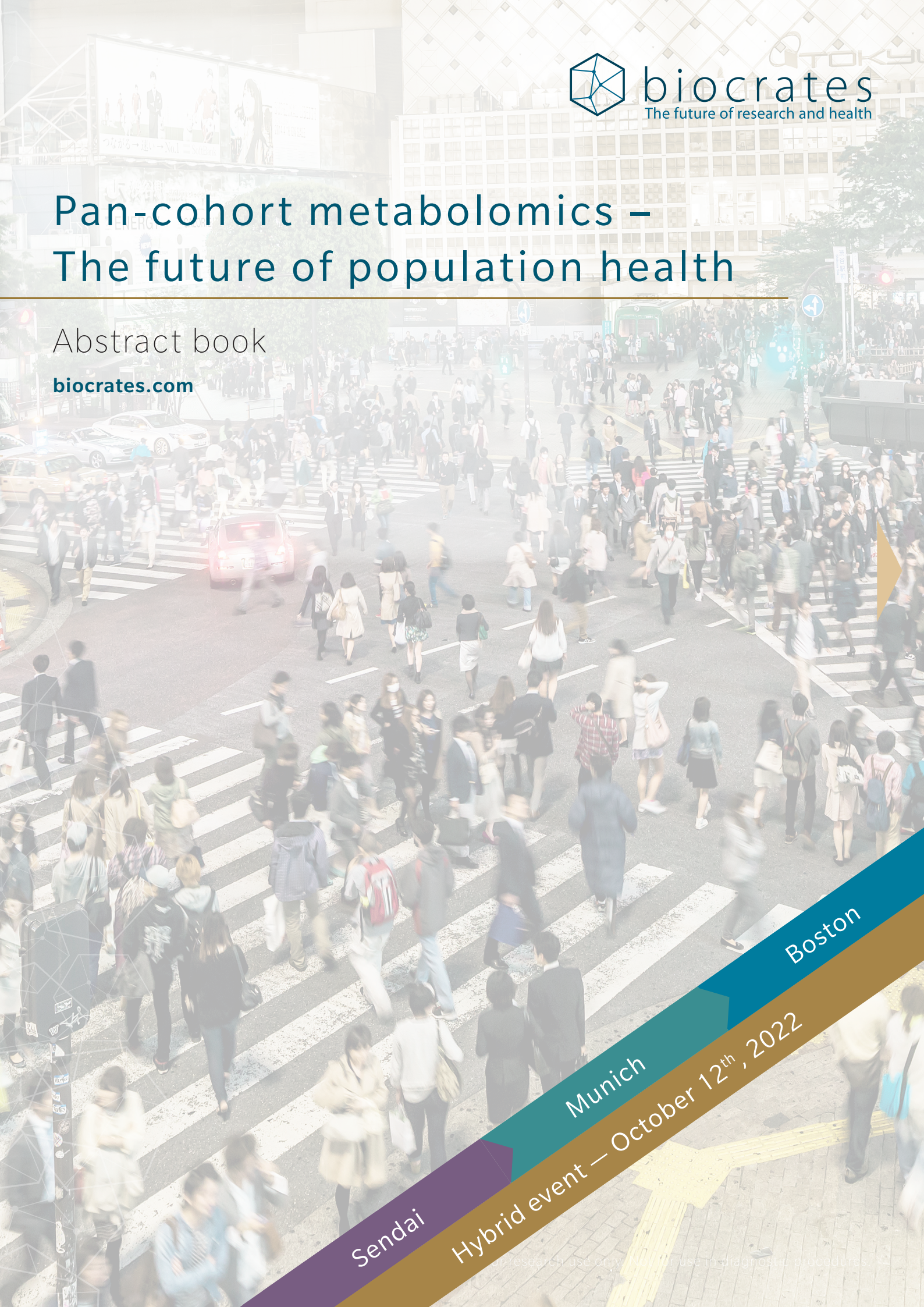

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TOHOKU MEDICAL MEGABANK ORGANIZATION

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Metabolomics – Enabling Precision Medicine
Prof. Rima Kaddurah-Daouk
Medicine and Psychiatry, Duke University,
Durham NC | USA
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Prof. Dr. Leroy Hood.
CEO | Founder, Phenome Health,
Seattle | USA
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The impact of population-based studies on population health
Prof. Jessica Lasky-Su
Brigham and Women's Hospital and Harvard Medical School,
Boston | USA
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Prof. Hitoshi Nakagama
President of the National Cancer Center, Tokyo | Japan
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Director of the joint Charité Universitätsmedizin Berlin and Berlin Institute of Health (BIH) | Germany
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Integrated Biobank of Tohoku Medical Megabank and space mouse study
Prof. Masayuki Yamamoto
Tohoku University Tohoku Medical Megabank Organization
Tohoku University, Sendai | Japan
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Session – Biobanks | Population-based studies

UK Biobank: A unique global study for health-related research and discovery science

Prof. Naomi Allen
Chief Scientist for UK Biobank, Oxford | United Kingdom

Singapore National Precision Medicine Program

Prof. John Chambers
Lee Kong Chian School of Medicine, Nanyang Technological University | Singapore

Taiwan Biobank resources for population health research

Chang Chung-ke, Ph.D.
Chief Administrative Officer and Chief Information Security Officer of Taiwan Biobank, Taipei | Taiwan

EPIC: A pan-European Cohort for the discovery of the causes of cancer and other chronic diseases

Marc Gunter, Ph.D.
Head of Nutrition and Metabolism Branch of the International Agency for Research on Cancer (IARC), Lyon | France

Tohoku Medical Megabank Project: Cohort and Biobank

Prof. Seizo Koshiba
Advanced Research Center for Innovations in Next-Generation Medicine, Tohoku University, Sendai | Japan

BioBank Japan: 20 years of disease-oriented cohort study towards genomic and multi-omic study

Prof. Takayuki Morisaki
BioBank Japan, The Institute of Medical Science, University of Tokyo | Japan

Germany: The cohort country

Prof. Henry Völzke
Institute for Community Medicine
University of Greifswald | Germany

Session – Precision medicine

Fast-tracking precision diagnostics/therapeutics for post-traumatic stress disorder & traumatic brain injury

Nicole Bjorklund, Ph.D.
Translational Research & Development Cohen Veterans Bioscience, New York | USA

Importance of the intestinal microbiota and metabolomic profiling in the balance between health and disease

Prof. Alessio Fasano
Center for Celiac Research and Treatment at Mass General Hospital for Children, Boston | USA

The future of population health: learning lessons from personalized healthcare

Prof. Alain van Gool
Personalized Healthcare of Radboud University Medical Center, Nijmegen | Netherlands

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Prof. Michael Knoflach
Center for Vascular Ageing & Stroke, Medical University of Innsbruck | Austria

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Prof. Siqi Liu
Chief Scientist Protein Sciences of BGI, Shenzhen | China

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Prof. Stefan Lorkowski
Nutritional Biochemistry University Jena | Germany

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Eugene Melamud, Ph.D.
Calico, San Francisco | USA

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Moritz von Scheidt, Ph.D.
Digimed Bavaria of the German Heart Centre Munich | Germany

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Prof. Michael Snyder
Genetics, School of Medicine | Stanford University | USA

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Prof. Claire Steves
Ageing and Health for King's College London | United Kingdom

Imaging metabolomics reveals roles of reactive sulfur species in pancreatic cancer chemoresistance

Prof. Makoto Suematsu
Department of Biochemistry, Keio University School of Medicine, Tokyo | Japan

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Prof. Toru Takebayashi
Preventive Medicine and Public Health School of Medicine for Keio University, Tokyo | Japan

Session – Omics data science

Privacy-preserving omics in medicine

Prof. Jan Baumbach
Institute for Computational Systems Biology, University Hamburg | Germany

Large-scale cohort study using targeted metabolomics kit and its application to gynecological cancer biomarker discovery

Ass. Prof. Eiji Hishinuma
Advanced Research Center for Innovations in the Next-Generation Medicine, Tohoku University, Sendai | Japan

Evotec E.MPD – Translational molecular patient database

Christiane Honisch, Ph.D.
SVP Diagnostics Devision | Evotec, Hamburg | Germany

Plasma biomarker studies on cancer within the Japan Public Health Center-based prospective study

Motoki Iwasaki, Ph.D.
Director of Epidemiology Research for the National Cancer Center, Tokyo | Japan

From multi-omics associations to molecular networks — making big results accessible

Gabi Kastenmüller, Ph.D.
Institute of Computational Biology, Helmholtz Munich | Germany

Pan-Cohort multi-omic signatures in health and disease

Prof. Jan Krumsiek
Institute for Computational Biomedicine, Weill Cornell Medicine, New York | USA

Expanding our genetic resources beyond European populations

Melissa Miller, Ph.D.
Human Genetics, Pfizer, Boston | USA

Statistical genetics, disease biology, drug discovery, and personalized medicine

Prof. Yukinori Okada
Department of Statistical Genetics, Osaka University Graduate School of Medicine | Japan

Insights from large-scale, multi-omic cohorts

Maik Pietzner, Ph.D.
Computational Medicine, Berlin Institute of Health at Charité – Universitätsmedizin Berlin | Germany

mGWAS: A powerful approach for metabolic fine mapping of genetic risk loci

Prof. Karsten Suhre
Director of Bioinformatics Core of Weill Cornell Medicine, Doha | Qatar

Pan-cancer analysis of pre-diagnostic blood metabolite concentrations

Vivian Viallon, Ph.D.
Nutrition and Metabolomics Branch of the International Agency for Research on Cancer (IARC), Lyon | France

Proteomics at population scale: insights from the UK biobank pharma proteomics project

Chris Whelan, Ph.D.
Data Science for Neuropsychiatry, Janssen, Boston | USA

Session – Omics technology

Study design for metabolomics research: challenges in biomedicine

Prof. Jerzy Adamski
Chief Scientific Officer of Metaron Diagnostics, Munich | Germany,
National University of Singapore | Singapore

Exposomics: measuring the environmental drivers of disease

Prof. Gary Miller
Environmental Health Sciences, Columbia University, New York | USA

Why targeted metabolomics is essential for population health

Prof. David Wishart
The Metabolomics Innovation Centre (TMIC) University of Alberta, Edmonton | Canada

BRAINCommons: A research and discovery platform paving the way for pan-cohort analysis

Maryan Zirkle, Ph.D.
BRAIN Commons of Cohen Veterans Bioscience, Edgewater | USA

Introduction

In recent decades, there has been an impressive increase in life expectancy across many regions of the world. Yet, along with declining rates of infectious disease mortality, there has been a considerable increase in the incidence and burden of chronic diseases. While global rates follow a uniform trend, there are striking regional differences that are speculated to result from different genetic backgrounds, lifestyles, nutrition, and other factors. However, additional studies are needed to clarify the roles of these factors in the prevalence of chronic diseases.

Studies addressing chronic diseases at the population level require a novel research approach. The approach must lead to a better understanding of disease pathology on the population scale while simultaneously enabling the delivery of personalized medicines. These requirements have prompted researchers to establish cohort studies in which millions of valuable biological samples have been collected from participants around the world and analyzed with various methods. At the same time, recent advances in omics technologies have revealed the tremendous opportunity to generate a comprehensive dataset from these samples when integrated with the existing epidemiological and clinical data. This dataset can ultimately help improve population health by personalizing disease prevention and treatment.

However, to maximize the value of omics within cohort studies, it is essential that researchers implement quality standards and establish international collaborations. This understanding prompted us to connect science globally with the symposium, "Pan-cohort studies – The future of population health." With this symposium, we sought to bring together researchers specializing in various aspects of cohort studies and biobanking. More than 40 speakers from academia and industry have contributed to more than 20 hours of scientific lectures and discussions over three overlapping meetings in Europe, North America and Asia. The speakers presented the concepts and results of multiple national and international population-based cohort studies and discussed common challenges for epidemiological studies. In particular, the presentations highlighted the challenges unique to collaboration between cohorts, such as data protection, participant compliance, interoperability of data, and validation of findings from independent cohorts. The speakers also shared insights on solutions, strategies, and tools to convert data into meaningful, actionable results. Moreover, they demonstrated the ability to translate findings to the clinic and impact precision medicine by implementing such solutions.

The discoveries and methodological foundations established in this symposium benefit many researchers. The findings not only aid researchers who aim to characterize epidemiological and clinical cohorts with metabolomics, but also pave the way for its translation into precision medicine approaches for disease prevention, clinical care, and treatment regimens. The symposium also demonstrated the importance of collaboration within epidemiological studies to facilitate advances in personalized medicine and improved population health.

We hope this symposium inspired innovative research ideas and new opportunities to collaborate. We are looking forward to continuing our work together, using metabolomics and multi-omics analysis to advance our understanding of the biochemistry of health and disease.

From the organizing team,

Dr. Therese Koal
Chief Technology Officer

Dr. Matthias Scheffler
Chief Business Officer,
Chief Scientific Officer

Metabolomics – Enabling Precision Medicine

Prof. Rima Kaddurah-Daouk

School of Medicine | Duke University,
Durham | USA

Abstract

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Beyond the Human Genome: A million person precision population health project that creates a foundation for the science of wellness and prevention

Prof. Dr. Leroy Hood

CEO of Phenome Health and CSO | Institute of Systems Biology, Seattle | USA

Abstract

The vision of this project is that we will develop the infrastructure to employ a data-driven (genome/phenome analyses) approach to optimizing the health trajectory of individuals for body and brain. We have two large populations (5000 and 10,000) that have respectively validated this approach for body and brain health, respectively. These studies have led to us pioneering the science of wellness and prevention as I will discussed in the lecture.

This project has required the acquisition of key partners for execution which will be necessary. We are approaching the Federal Government for funding for this project, as we did for the first Human Genome Project. This project is one of perhaps 10 or so 500,000 to one million person projects world-wide and it is unique in that it will carry out longitudinal phenome analyses, it will return results to participants and it is creating the infrastructure to spread this approach across the US and world healthcare systems. This project will lead to a powerful data ecosystem that will generate new knowledge about medicine, will catalyze the initiation of many start-up companies and will catalyze a paradigm shift in healthcare from its current disease orientation to a wellness and prevention orientation. This effort will catalyze the largest paradigm shift in medicine ever.

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The impact of population-based studies on population health

Prof. Jessica Lasky-Su

Harvard Brigham and Women's Hospital and Harvard Medical School, Boston | USA

Abstract

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Collaboration and integration of population-based and disease-based cohort studies are essential to the establishment of precision cancer medicine and personalized prevention

Prof. Hitoshi Nakagama

President of the National Cancer Center,
Tokyo | Japan

Abstract

Population-based cohort studies provide the best level of observational evidence on disease causation. They are especially powerful when a clinical trial is infeasible or when assessing multiple exposures or outcomes. Beginning in the 1980-90s, a number of large-scale population-based cohort studies have been established in Japan. Currently, these cohort studies have follow-up times of 20-30 years, and most have reached the fruitful period in which they are able to yield powerful epidemiological evidence of major disease outcomes, including cancer.
The Japan Public Health Center-based Prospective Study (JPHC Study), launched in 1990, includes around 140,000 residents aged 40-69 years across Japan who have provided information on lifestyle habits and health conditions in multiple follow-up surveys. The JPHC Study is among the first studies to have collected individual questionnaire information along with plasma and DNA samples, and clinical outcomes of multiple diseases. This study has reported associations between potential etiologic factors and the incidence of or mortality from cancer, and also other diseases associated with a relatively shorter life expectancy. In addition, the JPHC Study has reported associations between genetic/environmental factors and various diseases, and identified biomarkers that could be useful in predicting disease risk in individuals. Such scientific approaches at the population level have led Japan to help establish current "evidence-based" health policy at the national level. Various biomarker-based approaches have also been applied in population-based cohort studies. Deciphering associations between environmental exposures (e.g. smoking, etc) and mutational signatures is a good example. This kind of association is initially reported in patient cohorts, and then validated in general population cohorts, leading to the development of novel biomarkers for early diagnosis and behavioral changes aimed at avoiding cancer. Elucidation of epidemiological evidence from patient cohorts will likely be helpful in stratification of patients and precision medicine. Additionally, application and integration of such evidence into general population cohorts will also likely be critical to achieving the development of personalized cancer prevention.

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The role of cohort studies in prevention and personalized medicine

Prof. Annette Peters

Institute of Epidemiology,
Helmholtz Zentrum München | Germany

Abstract

The comprehensive data sets, collected when conducting population-based/cohort studies provide significant insight to challenges and trends in population-health-related questions.

However, an even larger impact could be generated, if the collected health data were also available to their respective individual owners/subjects. At some point in his or her life, every person will become a patient and in need of their personal health data for the best-possible personal diagnosis and treatment.

Relevant health-related data comes from all kinds of different sources - clinical and analytical data/ reports as much as more general health-care data like vital parameters collected at general practitioners or even fitness wearables. Health data collection but most importantly data management by creating patient-centered data spaces is in our understanding an essential next step in shaping the future of personal and population health. We will introduce and discuss possible connections with patient centered data spaces and translational research approaches to pan-disease cohorts.

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Why we need population-based studies

Prof. Christof von Kalle

Director of the joint Charité Universitätsmedizin Berlin and Berlin Institute of Health (BIH) | Germany

Abstract

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Integrated Biobank of Tohoku Medical Megabank and space mouse study

Prof. Masayuki Yamamoto

Tohoku University Tohoku Medical Megabank Organization
Tohoku University, Sendai | Japan

Abstract

The Tohoku Medical Megabank Project (TMM) has been launched to accomplish creative reconstruction in the aftermath of the Great East Japan Earthquake and ensuing tsunami 2011. TMM aims to establish an integrated biobank on the basis of two prospective large-scale cohort studies. The integrated biobank of TMM storages both bio-specimens and genome-omics data generated in-house. The latter includes genome and metabolome big data. TMM will share both information data and samples with the research community to facilitate biomedical research and personalized health care. TMM believes that constructing the integrated biobank by way of large-scale genome cohort studies will be effective in establishing the personalized health care and medicine.

Space stresses, including microgravity and cosmic radiation, are known to evoke various health problems in our body. Salient examples in astronauts are skeletal muscle loss and osteoporosis. It has been suggested that transcription factor NRF2 may contribute to the response against elevated stresses during spaceflight. Therefore, we conducted MHU-3 project in collaboration with JAXA, which sent six Nrf2 knockout (NRF2 KO) mice and six wild type (WT) mice into space to delineate the roles NRF2 plays during and after spaceflight. These mice were housed in the Japanese Experiment Module "Kibo" in the International Space Station for 31 days. We have conduced comprehensive transcriptome and metabolome analyses for the mice.

The NRF2 activity is indeed induced by space travel and contributes to the response against space stresses. We have set up the integrated biobank for Space Life Sciences (ibSLS) to facilitate the use of space mouse data. To the best of our knowledge, this study is the first to successfully complete a round trip of gene-modified mouse to the space. This study opens an avenue for the "Decade of Space Mouse".

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UK Biobank: A unique global study for health-related research and discovery science

Prof. Naomi Allen

Chief Scientist for UK Biobank,
Oxford | United Kingdom

Abstract

With its unique combination of scale, depth, maturity and accessibility, UK Biobank is enabling researchers worldwide to perform innovative health-related research. This talk will provide key information about this landmark study, highlight recent and future enhancements to the resource, and new ways in which it is democratising access via the new cloud-based Research Analysis Platform.

UK Biobank is a prospective cohort study of 500,000 people that has integrated large-scale genomic data (in all participants) and deep phenotyping data (including lifestyle factors, physical measures and multi-modal imaging) with long-term follow-up of health outcomes through linkage to electronic health records. The recent addition of detailed genomic sequencing data plus large-scale metabolomic and proteomic data has created an even more powerful resource, which will enable a better understanding of disease biology and will support innovative drug development for more effective therapies.

To accommodate the rapid growth of the resource and to enable more researchers across the world to access these data without limitations of transferring, collating, storing, and accessing data at this scale, UK Biobank has launched an cloud-based Research Analysis Platform, democratising access to large-scale compute and novel technologies. The availability of financial research credits for early-career researchers and those from low-income and middle-income countries is further democratising access to this unique resource.
Ready access to the combination of in-depth genomic, imaging, and other health information from 500,000 UK Biobank participants is enabling researchers worldwide to advance discovery science and improve human health.

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Singapore National Precision Medicine Program

Prof. John Chambers

Lee Kong Chian School of Medicine,
Nanyang Technological University Singapore
Singapore

Abstract

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Taiwan Biobank resources for population health research

Chang Chung-ke, Ph.D.

Chief Administrative Officer and Chief Information Security Officer of Taiwan Biobank,
Taipei | Taiwan

Abstract

The Taiwan Biobank was established as a national infrastructure for biomedical research. It currently boasts a population cohort of over 170,000 participants complete with blood plasma and urine biospecimens, and results from physical measurements, lifestyle questionnaires and serum/urine biochemical tests.

The Taiwan Biobank also collects several types of omics data, including whole genome genotype, whole genome sequence, DNA methylation status, plasticizer metabolites and nuclear magnetic resonance-based metabolomics. In this talk, I shall describe some interesting metabolomic characteristics of our population cohort.

I shall also showcase a few examples where the vertical integration of metabolome, genome and phenome data provided new insights into the health of the Taiwanese population, including common diseases and cancer. I will close the presentation with a discussion about possible opportunities for future pan-cohort collaborations using Taiwan Biobank resources.

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EPIC: A pan-European Cohort for the discovery of the causes of cancer and other chronic diseases

Marc Gunter, Ph.D.

Head of Nutrition and Metabolism Branch of the International Agency for Research on Cancer (IARC), Lyon | France

Abstract

The European Prospective Investigation into Cancer (EPIC) is a pan-European cohort comprising more than 520,000 individuals enrolled from ten countries (Norway, Sweden, Denmark, United Kingdom, The Netherlands, Germany, France, Italy, Spain and Greece) who have been followed since the mid-to-late 1990s. To date, more than 80,000 participants have developed cancer making EPIC one of the world's largest cohorts for studying the aetiology of cancer.

Diagnoses of other chronic diseases including diabetes, cardiovascular diseases as well as neurological diseases such as ALS, Parkinson's and Alzheimer's diseases have also been recorded. The original focus of EPIC was to capture variation in diet and lifestyle across Europe and to understand potential links with cancer development. A such, detailed information was collected from participants on dietary habits, physical activity and anthropometry. Baseline blood samples were collected from approximately 350,000 participants and aliquots are stored at the International Agency for Research on Cancer (IARC) and at the local EPIC centres.

Since its inception EPIC has become an important international resource for the discovery of new aetiological markers and pathways as well as biomarkers for early detection of cancer. Metabolomics data has been generated on >10,000 participants and a growing proportion of the cohort have genomics, proteomics and other targeted biomarkers available. This presentation will provide an overview of the EPIC cohort summarizing available resources, major findings and ongoing projects with a focus on the application of metabolomics profiling to understand new causes of cancer.

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Tohoku Medical Megabank Project: Cohort and Biobank

Prof. Seizo Koshiba

Advanced Research Center for Innovations in Next-Generation Medicine, Tohoku University,
Sendai | Japan

Abstract

Tohoku Medical Megabank (TMM) Project conducts two prospective cohort studies for more than 150,000 individuals in Japan. One is the TMM Community-Based Cohort Study targeting adult residents, and the other is the TMM Birth and Three-Generation Cohort Study targeting pregnant women with their infants, husbands, and both grandparents. We have already finished the baseline surveys (2013-2017) and the first repeat surveys (2017-2021) and are now conducting the second repeat surveys from 2021.

During the surveys, we have been collecting many kinds of data (questionnaires [lifestyles, medical history, etc.], blood and physiological tests, MRI, etc.) and samples (plasma, serum, urine, etc.) from participants. The collected information and samples are stored to the TMM Biobank system, including the large scale sample storage system and the database system (dbTMM), and are distributed to academia and industry. TMM also conducts genome and omics analyses of the collected cohort samples.

For genome analysis, TMM has already finished DNA array analysis for almost all adult participants and are now conducting whole genome sequence (WGS) project for 100K participants (more than 50K WGS have already finished). For omics analysis, TMM is conducting many kinds of omics analysis; metabolome, transcriptome, methylome, and metagenome. Especially, we have already finished plasma metabolome analysis for more than 50K participants. All obtained genome and omics data are also stored to our bio-bank system and are also distributed. Moreover, statistical information of our genome and omics data is freely available from our public database, Japanese Multi-Omics Reference Panel (jMorp). I will talk recent advances of our TMM project.

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BioBank Japan: 20 years of disease-oriented cohort study towards genomic and multi-omic study

Prof. Takayuki Morisaki

BioBank Japan, The Institute of Medical Science, University of Tokyo | Japan

Abstract

In 2003, BioBank Japan (BBJ) started developing one of the world's largest disease biobanks, recruiting a total of about 270,000 patients representing 440,000 cases of 51 common diseases with 12 medical institutions located throughout Japan. BBJ has collected DNA, serum, medical records including yearly collection with their consent for genomic and clinical research.

These biological samples and data are widely distributed and used by researchers. Large-scale genomic analyses, omics analyses including whole genome sequencing and biomarker analyses have been performed. As a result, more than 600 papers based on BBJ's samples and information have been published in international scientific journals, presenting research findings concerning, for example, the association between genetic information of Japanese individuals and the onset of various diseases.

Discoveries have been reported of genes related to diseases and drugs as well as physical traits and biomarkers. Currently, BBJ is especially focusing on genomic and muli-omic study using genome/metabolome/proteome data.

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Germany: The cohort country

Prof. Henry Völzke

Institute for Community Medicine
University of Greifswald | Germany

Abstract

Since the 1980ies, multiple regional and national cohort studies have been established in Germany. The presentation will reflect German cohort research. Special focus will be laid on the German National Cohort (NAKO) and the Study of Health in Pomerania (SHIP), which can be regarded as major sources for biomarker research.

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Session – Precision medicine
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Fast-tracking precision diagnostics/ therapeutics for posttraumatic stress disorder and traumatic brain injury

Nicole Bjorklund, Ph.D.

Translational Research & Development | Cohen Veterans Bioscience, New York | USA

Abstract

Despite extensive research to characterize psychological, genomic, and physiological risk and etiologic factors, there are currently few validated biomarkers for Posttraumatic Stress Disorder (PTSD) and Traumatic Brain Injury (TBI). Many potential biomarkers of PTSD & TBI published in the literature have not been independently replicated or advanced through a qualification process for regulatory approval and use. Developing biomarker-based diagnostics is essential to shifting the diagnosis & treatment of PTSD and TBI from a syndromic, symptom-based approach to a biological, mechanistically-based one that targets the effects of trauma at their molecular roots.

Harnessing the advancements in multi-omics approaches along with cutting-edge data analytics can fast track the discovery and development of biomarker candidates. Cohen Veterans Bioscience (CVB), a 501(c)3 nonprofit biomedical research and technology organization has built critical infrastructure to support the development of biomarkers with the overarching goal of developing multi-modal mechanistic disease models. These resources include a biorepository housing specimens collected using best practices from well-characterized clinical cohorts, genomic & imaging analytics pipelines to support multi-omic approaches, BRAINCommons data sharing platform, and a fluid-based assay evaluation paradigm. A case-study of the approach will be presented along with results and takeaway lessons.

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Importance of the intestinal microbiota and metabolomic profiling in the balance between health and disease

Prof. Alessio Fasano

Center for Celiac Research and Treatment
at Mass General Hospital for Children,
Boston | USA

Abstract

Improved hygiene leading to a reduced exposure to microorganisms have been implicated as one possible cause for the recent 'epidemic' of chronic inflammatory diseases (CID) in industrialized countries. That is the essence of the hygiene hypothesis that argues that rising incidence of CID may be, at least in part, the result of lifestyle and environmental changes that have made us too "clean" for our own good.

The gut microbiome consists of more than 100 trillion microorganisms, mostly bacteria. It has been just recently recognized that there is a close bidirectional interaction between gut microbiome and our immune system, and this cross talk is highly influential in shaping the host gut immune system function and, ultimately, shifting genetic predisposition to clinical outcome.

This observation led to a revisitation of the possible causes of CID epidemics, suggesting a key pathogenic role of microbiome composition. However, to proper interpret the impact of microbiome composition and function in disease pathogenesis, prospective studies integrating metagenomic data with subjects' genomic, metadata, and metabolomic profiling are necessary. This multi-omic analysis will be instrumental to develop strategies for personalized interventions (precision medicine) and even disease interception (primary prevention).

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Session – Precision medicine
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The future of population health: learning lessons from personalized healthcare

Prof. Alain van Gool

Personalized Healthcare of Radboud University Medical Center, Nijmegen | Netherlands

Abstract

We have reached a fantastic period in biomedical science. Exponential developments in molecular laboratory technologies such as next generation sequencing and mass spectrometry have enabled us to obtain increasing insights in the molecular components of human biology and their interactions. Novel personalized diagnostics and high precision therapies that modulate selected disease mechanisms are now driving the new paradigm of precision medicine. The parallel strong developments in digital biomarker platforms like wearables and apps further drive the personalized aspect of health management, even towards prevention of un-health. Collectively, we now aim to translate interdisciplinary research to knowledge, understanding and actionable decisions for people to maintain and/or improve health, both at the population as on the individual level.

However, while embarking on the road towards precision medicine and health, we are rediscovering that human physiology in combination with environmental factors is a highly complex system and that we need multiple viewing angles to begin to understand the complexity, identify its key nodes and define optimal therapeutic approaches. To innovate to the next level, we need to be fully aware of the many lessons learned thus far in population health and personalized health, and use these insights to translate novel capabilities to daily practice.

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Lessons learned from large clinical cohorts — Experience from the VASCage consortium

Prof. Michael Knoflach

Center for Vascular Ageing & Stroke |
Medical University of Innsbruck
Innsbruck | Austria

Abstract

VASCage is Competence Centers for Excellent Technologies (COMET) funded by the Austrian Research Promotion Agency focusing on vascular ageing and stroke. One core element of this research company is the central biobank with collection of biosamples with detailed clinical annotations. This talk will give brief overview over problems, pitfalls and strengths of conducting clinical cohort research.

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Session – Precision medicine
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Evaluation of spike protein epitopes by assessing dynamics of the humoral immune responses in the moderate COVID-19

Prof. Siqi Liu

Chief Scientist Protein Sciences of BGI,
Shenzhen | China

Abstract

The coronavirus disease 2019 (COVID-19) pandemic is caused by a novel coronavirus called severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2). The spike protein (S) of SARS-CoV-2 is a major target for diagnosis and vaccine development because of its essential role in viral infection and host immunity.

Currently, the time-dependent responses of humoral immune system against various S protein epitopes are poorly understood. In this study, enzyme-linked immunosorbent assay (ELISA), peptide microarray, and antibody binding epitope mapping (AbMap) techniques were used to systematically analyse dynamic changes in the humoral immune responses against S protein in a small cohort of moderate COVID-19 patients that were hospitalized for approximately 2 months after the onset of symptoms.

The recombinant truncated S proteins, target S peptides and random peptides were taken as antigens in the analyses. The assays demonstrated dynamic IgM- and IgG recognition against various S protein epitopes appearing patient-dependent patterns. Comprehensive analysis of epitope distribution along the Spike gene sequence and spatial structure of the homotrimer S protein demonstrated that most IgM- and IgG-reactive peptides were clustered in the accessible regions of accessible regions of the S1, S2 and RBD domains. Seven S peptides were recognized by the IgGs derived from the serum samples of all COVID-19 patients. The dynamic immune recognition signals from these seven S peptides were comparable to the entire S protein or the truncated S1 protein. Furthermore, in this cohort, individual patients demonstrated stable immune recognition to certain S protein epitopes throughout their hospitalization period. The dynamic characteristic of the humoral immune responses to S protein, therefore, has provided valuable information for further design of diagnosis and immunotherapy of COVID-19.

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Open data for science — A personal note on hurdles and pitfalls

Prof. Stefan Lorkowski

Nutritional Biochemistry University Jena,
Jena | Germany

Abstract

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Session – Precision medicine
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Predicting longevity — integrative modeling of plasma biomarkers and imaging across disease space

Eugene Melamud, Ph.D.

Calico Life Sciences,
San Francisco | USA

Abstract

There is a multitude of pathological conditions that affect human health, yet we currently lack a predictive model for most diseases, and underlying mechanisms that are shared by multiple diseases are poorly understoodIn this talk, we will present an epidemiological analysis of large-scale biomarker and imaging datasets to identify the most important factors prognostic of future disease and mortality.

We will show how proximity in the biomarker-disease space is strongly related to the occurrence of disease comorbidity, suggesting biomarker profile patterns can be used for both predicting future outcomes as well as a sensitive mechanism for detecting under-diagnosed disease states. We will further show that maintenance of calcium/phosphate homeostasis is a broad contributor to human aging rate and longevity.

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DigiMed Bayern — digitized, personalized medicine in atherosclerosis

Moritz von Scheidt, Ph.D.

Digimed Bavaria of the German Heart Centre,
Munich | Germany

Abstract

Abstract not available.

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Session – Precision medicine
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Big data, health and COVID19

Prof. Michael Snyder

Genetics, School of Medicine | Stanford University, Stanford | USA

Abstract

Recent technological advances as well as longitudinal monitoring not only have the potential to improve the treatment of disease (Precision Medicine) but also empower people to stay healthy (Precision Health). We have been using advanced multiomics technologies (genomics, immunomics, transcriptomics, proteomics, metabolomics, microbiomics) as well as wearables for monitoring health in 109 individuals for up to 12 years and made numerous major health discoveries covering cardiovascular disease, oncology, metabolic health and infectious disease.

We have found that individuals have distinct aging patterns that can be measured in an actionable period of time as well as seasonal patterns of health markers. We have also explored the effects of fiber using multiomics profiling and profile dynamics during pregnancy. Finally, we have used wearable devices for early detection of infectious disease, including COVID-19 and built an alerting system for detecting health stressors that is scalable to the entire planet. We believe that advanced technologies have the potential to transform healthcare.

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The future of research and health

Cohort studies in the digital era: Use of time-series tracking and experimentation in TwinsUK and beyond

Prof. Claire Steves

Ageing and Health for King's College London
London | United Kingdom

Abstract

TwinsUK is one of the most deeply phenotyped and assayed twin cohorts in the world, with longitudinal data collection going back 30 years. TwinsUK data initially contributed to genetic consortia and was the seventh most used datasets in the world. TwinsUK has then pioneered the use of many omics in relation to epidemiology of ageing traits and diseases, including epigenetics, transcriptomics and metabolomics. Over the last 5 years we have used time-series of clinical and biological data in response to experimental challenges to bring new insights into vulnerability to disease.

Today I will describe some of these dynamic phenotyping studies, including the Predict study, and how this led to the capability to measure time-series data on a massive scale, across the UK over the COVID-19 pandemic. This work on the COVID symptoms Study, in collaboration with ZOE, led to several world firsts in the understanding of the disease. Within the TwinsUK cohort, during the COVID pandemic we have used novel approaches to capture the social, psychological and biological impact of both COVID itself, and measures used to address it, which will form the basis of collaborative work on the long term impact on health and wellbeing. As well as using such natural 'experiments' we have demonstrated that experiments within the cohort setting can add unique insight to the major challenges of our time.

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Session – Precision medicine
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Imaging metabolomics reveals roles of reactive sulfur species in pancreatic cancer chemo-resistance

Prof. Makoto Suematsu

Department of Biochemistry, Keio University School of Medicine, Tokyo | Japan

Abstract

Formalin-fixed-paraffin-embedded (FFPE) tissue defaults to a standard to diagnose malignancy but is inappropriate for detecting marker metabolites in situ. Post-operative frozen samples of pancreatic ductal adenocarcinoma (PDAC) were analyzed by imaging metabolomics (IM), imaging mass-spectroscopy and gold-nanoparticle-based SERS imaging, which cover large areas of cancer and stromal regions.

IM shows that polysulfide occurs in all regions concurrently with stromal enrichment of cystine, a substrate of reactive sulfur species. In FFPE samples from 120 PDAC patients, immuno-staining polysulfide-generating enzymes revealed that cystathionine gamma-lyase (CSE) expressed in cancer-associated fibroblasts (CAF) serves as an independent factor worsening post-operative disease-free and overall survivals. Polysulfide renders cancer cells to induce fascin-actin-bundling protein-1 to stimulate the cancer cell motility. These results suggest that polysulfide serves as a marker inducing CAF-mediated cancer cell activation.

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Metabolomics and cardiometabolic risk: Findings from the international collaborative analysis

Prof. Toru Takebayashi

Preventive Medicine and Public Health School of Medicine for Keio University,
Tokyo | Japan

Abstract

Burden of non-communicable diseases differs country-by-country, and various aspects such as environmental, cultural and genetic factors could explain these differences. This is true for metabolite profiles. International collaborative analysis of metabolomics epidemiology can contribute to find out common and/or unique metabolites/metabolic pathways reflecting genetic and/or cultural backgrounds. We initiated Tsuruoka Metabolomics Cohort Study (TMCS) enrolling 11,002 community-dwelling adults in Tsuruoka City of Japan in 2012, in which biospecimen sampling and analytical procedures has been standardized and optimized for metabolomics as a large-scale cohort. Plasma and urinary metabolites were quantified by CE/MS for charged metabolites and LC/MS for others at baseline and thereafter. Reproducibility and validity of our metabolite measurements for both plasma and urine have been confirmed using QC samples.
Two comparative studies with Baltimore Longitudinal Study of Aging (BLSA) and TMCS have been done to explore blood metabolite signature of metabolic syndrome (MetS) using our CE/MS (study 1) and biocrates platform (study 2). In study 1, we identified 18 metabolites shared between TMCS and BLSA among top 25 most significant metabolites in each cohort, and the majority of which were classified as amino acids including branched-chain amino acid metabolism, glutathione production, aromatic amino acid metabolism, gluconeogenesis, and the tricarboxylic acid cycle. In study2, we found that metabolites from the phosphatidylcholines-acyl-alkyl, sphingomyelin, and hexose classes were significantly associated with MetS and risk factor outcomes in both cohorts. TMSC is also a member of COMETs, and the result of an international pooled analysis of circulating trimethylamine N-oxide (TMAO) of 16 population-based studies indicated the associations of circulating TMAO with high intakes of animal protein across populations and that with multiple cardiometabolic risk factors, including impaired renal function and poor glycemic control. We will discuss what the commonalties and differences across cross-ethnic/cultural comparison we observed were and how to move international collaboration across metabolomics cohorts forward.

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Session – Omics data science
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Privacy-preserving omics in medicine

Prof. Jan Baumbach

Institute for Computational Systems Biology,
University Hamburg | Germany

Abstract

Large-scale omics data analyzed by artificial intelligence (AI) technology is finding its way into the clinic to revolutionize our approach to medicine as a whole. Beyond personalized medicine, AI-driven medical data profiling is leaving massive footprints – from drug repurposing to a mechanistic redefinition of diseases. To move away from organ- and symptom-based disease descriptors to clinically actionable mechanistic approaches, computational systems and network medicine emerged.

We will introduce the field, discuss current approaches and pitfalls as well as potential future avenues. Finally, we will introduce privacy-preserving AI and machine learning technology working on distributed medical data to extract predictive systems medicine profiles, specifically from highly sensitive genomics and transcriptomics data.

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Large-scale cohort study using targeted metabolomics kit and its application to gynecological cancer biomarker discovery

Ass. Prof. Eiji Hishinuma

Advanced Research Center for Innovations in the Next-Generation Medicine | Tohoku University,
Sendai | Japan

Abstract

Metabolomics, a method for comprehensive analysis of metabolites in vivo, is considered to reflect human phenotypic variation relatively well and has contributed to biomarker discovery research. However, there have been very few examples of its application to clinical practice. Therefore, it is very important to clarify in vivo metabolome profiles by utilizing samples accumulated in large-scale cohort studies for the identification of disease biomarkers.

The Tohoku University Tohoku Medical Megabank Organization (ToMMo) collects a wide variety of samples from large-scale cohort studies and stores them in a biobank, where the plasma samples are used for metabolome analysis. To date, plasma metabolome analyses using the targeted metabolomics kit have been performed on several thousand individuals, and the results are published in ToMMo's database, the Japanese Multilayer Omics Reference Panel (jMorp). Recently, the Tohoku University Clinical Biobank was established, and specimens derived from patients at Tohoku University Hospital have also been collected, and metabolomic analysis of these specimens is also being conducted. In this presentation, we describe the results of a study on the analysis of ToMMo plasma samples using the targeted metabolomics kit and the evaluation of the quantitative values obtained.

In addition, we present the results of a comparison of gynecological cancer samples from the Clinical Biobank with cohort samples to identify differences in plasma metabolomic profiles and biomarkers associated with disease prognosis. Our research demonstrated that plasma metabolome analysis is useful not only for cancer diagnosis, but also for predicting prognosis and evaluating of chemotherapy response with the variation of metabolites.

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Session – Omics data science
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Evotec E.MPD – Translational molecular patient database

Christiane Honisch, Ph.D.

SVP Diagnostics Devision | Evotec, Hamburg | Germany

Abstract

Abstract not available.

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The future of research and health

Plasma biomarker studies on cancer within the Japan Public Health Center-based prospective study

Motoki Iwasaki, Ph.D.

Director of Epidemiology Research for the National Cancer Center, Tokyo | Japan

Abstract

The Japan Public Health Center-based prospective Study (JPHC study) conducted a baseline survey for 140 420 registered residents aged 40–69 years within 11 public health center areas nationwide in 1990–94.

Five- and 10-year follow-up surveys were conducted to update information on lifestyle habits and health conditions. About 130 000 participants responded to at least one of the three questionnaire surveys and about 60 000 participants provided blood samples on at least one of the two sampling times. The subjects have been followed for vital status and the occurrence of cancer and other diseases.

As of October 2021, 44 000 deaths have been documented, as well as 32 000 cases of cancer. In order to examine the association between plasma biomarkers and the risk of cancer, we applied both nested case-control and case-cohort design to the JPHC study. In this symposium, I will present several findings from both designs as examples. I will discuss about utilizing plasma biomarkers for identifying risk or predictive factors and implication of these findings for cancer etiology and prevention.

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From multi-omics associations to molecular networks — making big results accessible

Gabi Kastenmüller, Ph.D.

Institute of Computational Biology, Helmholtz Munich, Munich | Germany

Abstract

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The future of research and health

Pan-cohort multi-omic signatures in health and disease

Prof. Jan Krumsiek

Institute for Computational Biomedicine, Weill Cornell Medicine, New York | USA

Abstract

In this presentation, I will discuss various research projects related to multi-omic signatures spanning across population and patient cohorts. (1) We will talk about how variational autoencoders extract universal metabolomic components from the TwinsUK cohort, and how these components correlate with disease parameters in independent multi-ethnic disease cohorts. (2) In several COVID-19 related projects, we used multi-omics data to gain insights into the complicated pathophysiology of this novel disease and develop models for the prediction of outcome severity in the hospital. (3)

In Alzheimer's d disease projects, we are integrating data from brain and the periphery for a better understanding of the disease and the development of novel therapeutic leads. (4) We have assembled a combined metabolomic/transcriptomic cohort of over 1,000 cancer tissue samples, which provide deep insights into the co-regulation of these two omics layers. (5) Finally, I will present an outlook on comparative oncology, where we perform an integrated metabolomic analysis of human- and dog-based lymphoma data.

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Expanding our genetic resources beyond European populations

Melissa Miller, Ph.D.

Human Genetics, Pfizer,
Boston | USA

Abstract

Large-scale population biobanks linked to genetic data have proved to be indispensable resources in the genetics community, enabling genetic discovery for diseases and phenotypes across a wide-spectrum of human biology. Many of the large population biobanks are pre-dominantly or exclusively of European populations.

The lack of diversity in genetics limits has several downstream implications. Most simply, this limits ability for discovery of novel genetic associations that may not be present in European populations. Lack of diversity in genetics can also lead to lack of translatability as many polygenic risk scores and precision medicine discoveries do not translate well to non-European populations. Taken together, these biases can exacerbate disparities in healthcare.

In recent years, multiple groups have started building biobanks and genetic cohorts in non-European populations (Mexican biobank, example in Africa). These efforts have already started to provide insight into genetic diversity in other regions of the world. More importantly, many of these new efforts have been developed as partnerships with an explicit goal of building resources and infrastructure in their countries and continents.

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The future of research and health

Statistical genetics, disease biology, drug discovery, and personalized medicine

Prof. Yukinori Okada

Department of Statistical Genetics, Osaka University
Graduate School of Medicine
Osaka | Japan

Abstract

Statistical genetics is a research field that evaluates causality of human genetic variations on diseases, using statistical and bioinformatics approaches. Recent developments of sequencing technologies have provided human disease genome data of hundreds of thousands of the subjects, and successfully identified comprehensive catalogues of genetic susceptible loci.

However, little is known regarding how to develop methodology to integrate large-scale human genome data with diverse biological resources. We have developed such methods and applied to a pioneering example of large-scale genetic association studies on a variety of human complex traits. Tran-layer omics analysis identified the cell types and microbiomes implicated in disease biology. Network analysis between the disease risk genes and the drug target genes could identify novel candidates of drug repositioning.

Integration of cell type-specific gene expression profiles estimated from GWAS with compound perturbation databases can pinpoint novel therapeutic targets and compounds. Application of the machine learning methods into population genome data can classify the samples without prior biological information. These results should empirically show the value of statistical genetics to dissect disease biology, novel drug discovery, and personalized medicine. Finally, we would like to introduce our activity on young researcher developments (“Summer school of statistical genetics” in Osaka Univeristy).

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Session – Omics data science
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Insights from large-scale, multi-omic cohorts

Maik Pietzner, Ph.D.

Computational Medicine, Berlin Institute of Health at Charité – Universitätsmedizin Berlin,
Berlin | Germany

Abstract

Profiling small molecules circulating in blood at scale allows unprecedented insights in disease aetiology when done in large prospective cohorts and coupled with DNA sequence variation. In this talk I will give an overview about our recent work linking about 1000 metabolites to the incidence of 27 diverse diseases spanning multiple clinical specialties. I will demonstrate, that while we see hundreds of metabolites to be prospectively associated with disease risk and all-cause mortality there is little specificity in metabolite-disease associations with more than two-thirds of metabolites associated with two or more diseases.

We traced these finding back to common antecedents of multimorbidity, the presence of multiple chronic diseases in the same patient. In the second part of the talk, I will provide a brief dive into the genetic determinants of blood metabolite levels among >20,000 participants, to finally address the questions whether metabolite – disease associations are cause or consequence by presenting a phenome-wide 'genetic metabolic load' study in more than 300,000 participants.

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mGWAS: A powerful approach for metabolic fine mapping of genetic risk loci

Prof. Karsten Suhre

Director of Bioinformatics Core of Weill Cornell Medicine,
Doha | Qatar

Abstract

In the last 20 years genome wide association studies (GWAS) have discovered a great number of associations between genetic variance and (clinical) phenotype. Extending this concept with metabolomics (Mx) data into mGWAS leads to a powerful, but still undervalued, approach to qualify genomic variance by druggability (phenotype converting effect size) and to shift from association to functional relationship.
Starting in 2008 with an mGWAS in merely 300 individuals (Gieger et al., PLoS Genetics, 2008), the approach matured with over 80 mGWAS studies published to date: http://www.metabolomix.com/list-of-all-published-gwas-with-metabolomics/. As of 2022, genomic (Gx) and metabolomic data at the scale of large population-based cohort are available, with over 100,000 samples from the UK biobank based on Gx and NMR-based Mx data.

In this presentation we demonstrate that Mx association profiles can serve as surrogate endpoints for future drug trials and Mendelian randomization studies, as shown for lipid-lowering treatment and incident myocardial infarction, pointing to a target with currently unexploited therapeutic potential.

Mx association profiles from mGWAS provide a resource for the functional interpretation of lipid risk loci and their evaluation as potential drug targets in the prevention and treatment of ASCVD.

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Pan-cancer analysis of pre-diagnostic blood metabolite concentrations

Vivian Viallon, Ph.D.

Nutrition and Metabolomics Branch of the International Agency for Research on Cancer (IARC),
Lyon | France

Abstract

Background: Epidemiological studies of associations between metabolites and cancer risk have typically focused on specific cancer types separately. Here, by leveraging metabolomics data available in a large international multi-centric cohort study, we designed a multivariate pan-cancer analysis to identify metabolites potentially associated with multiple cancer types, while also allowing the investigation of cancer type-specific associations .

Methods: We analyzed targeted metabolomics data available for 5,828 matched case-control pairs from cancer-specific case-control studies on breast, colorectal, endometrial, gallbladder, kidney, localized and advanced prostate cancer, and hepatocellular carcinoma nested within the European Prospective Investigation into Cancer and Nutrition (EPIC) cohort. From pre-diagnostic blood levels of an initial set of 117 metabolites, 33 cluster representatives of strongly correlated metabolites, and 17 single metabolites were derived by hierarchical clustering. The mutually adjusted associations of the resulting 50 metabolites with cancer risk were examined in penalized conditional logistic regression models adjusted for body mass index, using the data shared lasso penalty.
Results: Out of the 50 studied metabolites, (i) six were inversely associated with risk of most cancer types: glutamine, butyrylcarnitine, lysophosphatidylcholine a C18:2 and three clusters of phosphatidylcholines (PCs); (ii) three were positively associated with most cancer types: proline, decanoylcarnitine and one cluster of PCs; and (iii) 10 were specifically associated with particular cancer types, including histidine that was inversely associated with colorectal cancer risk, and one cluster of sphingomyelins that was inversely associated with risk of hepatocellular carcinoma and positively with endometrial cancer risk.

Conclusions: These results could provide novel insights for the identification of pathways for cancer development, in particular those shared across different cancer types.

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Proteomics at population scale: insights from the UK biobank pharma proteomics project

Chris Whelan, Ph.D.

Data Science for Neuropsychiatry | Janssen, Boston | USA

Abstract

This talk will provide an overview of the rationale for conducting high-throughput proteomics at population scale, including recent learnings from multiplex proteomic profiling of 54,306 individuals in the UK Biobank.

No recording available

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Study design for metabolomics research: challenges in biomedicine

Prof. Jerzy Adamski

Chief Scientific Officer of Metaron Diagnostics,
Munich | Germany, National University of Singapore | Singapore

Abstract

Only robust and validated biomarkers are of interest in the diagnostics and theranostics. Metabolomics research facilitates development of biomarkers for precision medicine.

During a study design there are several specific requirements which are a prerequisite for successful study in biomedicine. Critical components include power calculation, consideration of confounders, definition of controls or references, aspects of ethnicity and environmental impact, further sample randomization for analytics and the choice of analytical methods.

These interlaced components if not included may cause severe overfitting and even loss of association with phenotype.

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biocrates
The future of research and health

Exposomics: Measuring the environmental drivers of disease

Prof. Gary Miller

Environmental Health Sciences, Columbia University,
New York | USA

Abstract

Advances in high-resolution mass spectrometry (HRMS) have led to an increased ability to measure small molecules in biological samples. Although the field of metabolomics has progressed in its ability to measure endogenous small molecules, little attention has been given to the detection of environmental chemicals and other xenobiotics in human samples using these high-resolution approaches, which is termed exposomics.

Dr. Miller will discuss the approaches his team has taken to better understand the environmental drivers of disease. Several major research initiatives have been established to identify causes of human disease with massive biobanks created. Development of techniques that can provide omic-scale analysis of environmental chemicals in biobanked samples could rapidly increase our understanding of environmental influences of disease. Dr. Miller's group is focused on the use of liquid chromatography and gas chromatography-based high-resolution mass spectrometry to detect a wide range of xenobiotics and their metabolites. Along with colleagues in the EU-funded ESFRI project EIRENE, the team is working to develop an exposomic analysis framework for human clinical, translational, and population studies, which will allow seamless integration into multi-omic studies of human disease that can be used within large-scale efforts. Dr. Miller will discuss recent data from several studies that are underway.

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Session – Omics technology
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Why targeted metabolomics is essential for population health

Prof. David Wishart

The Metabolomics Innovation Centre (TMIC) University of Alberta, Edmonton | Canada

Abstract

In this presentation I will describe the benefits of using targeted, fully quantitative metabolomics for conducting population health studies. Metabolomics can be divided into two camps: untargeted and targeted approaches. Untargeted metabolomics emphasizes coverage over quantification. Targeted metabolomics emphasizes quantifcation over coverage. Currently, metabolomics is experiencing a disturbing shift towards untargeted methods and away from absolute quantification. This is dangerous. To indicate why this is dangerous, I will breifly describe the challenges that the field of proteomics faced when it moved completely towards untargeted approaches and the consequences that this "collective" decision had to the field.

To counter this story of caution, I will also describe some of the beneifts of performing absolute quantification in metabolomics. Specifically, I will show how quantification improves reproducibility, how targeted metabolomics improves speed and reduces costs and how targeted metabolomics often exceeds the coverage obtained by untargeted methods. I will also highlight how targeted metabolomics is required for ISO and CLIA certification, how the the discovery of biomarkers can be simplified with targeted metabolomics and why the translation of discoveries to the clinic and for populatio health studies is far easier with targeted metabolmics than untargeted metabolomics. I will briefly provide some examples of how we have used targeted metabolomics or the data from targeted metabolomics to impact population health.

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biocrates
The future of research and health

BRAINCommons: A research and discovery platform paving the way for pan-cohort analysis

Maryan Zirkle, Ph.D.

BRAIN Commons of Cohen Veterans Bioscience,
Edgewater | USA

Abstract

We are in a new era of data-driven biomedical informatics research, where organizations with powerful data acquisition technologies are generating high-quality, health data at increasing scales. Our primary challenge today is harnessing the combined power of the data for accelerated discovery and increased impact. Cohen Veterans Bioscience (CVB), a 501(c)3 nonprofit biomedical research and technology organization developed the BRAINCommons™ (BC), a next-generation, cloud-based research platform that enables data-driven discovery across disparate cohorts or datasets.

Leveraging interoperability approaches that harmonize content and format of data from multiple studies or projects, we increase the value, enabling the creation of new cohorts for additional analysis and discovery of meaningful insights. The BC is in the early stages of implementation, currently hosting 15 cohorts with more than 8,000 study participants with Parkinson's Disease (PD), Post-traumatic Stress Disorder (PTSD) and/or Traumatic Brain Injury (TBI) from US, Canada, Europe and beyond. CVB, a primary customer of the BC, sponsors several deep-phenotyping clinical studies, including a multi-site National Normative Neuro-imaging Library (NNL) study conducted at three Academic Medical Centers.

This study has a target enrollment of 3000 individuals with a full battery of brain MRI sequences, biosample collection, and over 25 different clinical assessments for each individual. In this multi-year study, data has already been collected from over 1400 individuals and the raw imaging files and clinical data are being housed in the BC.

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Flashtalks

A special thank you to all speakers of the Flashtalk sessions giving a short but comprehensive overview of their studies during the breaks of the live events.

A metabolomics study of stroke in REGARDS – Reasons for geographic and racial differences in stroke

Dr. Zsuzsanna Ament
Department of Neurology
Harvard Medical School, Boston | USA

The human serum metabolome – A combined LC-MS/MS and NMR approach to explore Parkinson´s disease and its associated biomarkers

Nadia Ashrafi
Metabolomics Research
Beaumont Health Royal Oak | USA

Standardized biobanking as a boost for personalized medicine

Dr. Anna Michalska-Falkowska
Medical University of Bialystok | Poland

Shortened leucocyte telomere length is associated with increased daily air temperature: KORA F3 and KORA F4

Dr. Wenli Ni
Institute of Epidemiology
Helmholtz Munich | Germany

Using pan-cohort data for discovery at Clinical Microbiomics

Dr. Henrik Bjørn Nielsen
Chief Scientific Officer,
Clinical microbiomics

Machine learning for classification of hypertension subtypes using multi-omics

Dr. Parminder Singh Reel
School of Medicine
University of Dundee | United Kingdom

Longitudinal associations between metabolites and long-term exposure to ambient air pollution: results from the KORA cohort study

Dr. Yueli Yao
Institute of Epidemiology,
Helmholtz Munich | Germany

Closing remarks

The symposium "Pan-cohort studies: the future of population health" was an inspiring day of research and discussion.

Reflecting on the event, we can take away that the samples from population-based cohort studies and biobanks are treasures waiting to be raised, as they comprise the information about how ethnicities, lifestyles, and exposition contribute to the rise of chronic diseases in an aging population. A couple of points strike us as essential for successful -omics research within cohort studies and collaboration between cohorts:

— Generation and integration of multi-omics data across population studies requires standardized solutions and procedures.
– To contribute significantly to population health, omics data must reflect human diversity. Even today, 80% of all collected genetic data is linked to Western European and North American populations. Therefore, it was great to learn about the work being conducted in regions such as Asia, South America, and Eastern Europe.
– Data accessibility for all stakeholders of the healthcare ecosystem is essential and to the benefit of us all – but the topic has to be addressed simultaneously with data security and privacy regulations.

All of us can contribute to making the best of sample collections through cohort studies and biobanks. Through formal collaborations and making data available to the scientific community, we can improve our understanding of the biochemical processes that define health and disease. At biocrates, we are committed to providing solutions that enable successful metabolomics and multi-omics research.

We are hugely thankful for everyone who has helped to make our meeting a success:

– Thanks to all 40 speakers for being so generous with their time and sharing their knowledge in their fantastic presentations.
– Thanks to our global audience for joining us on our 22h journey of connecting science globally.
– Thanks to our local hosts and session chairs for their contribution and commitment to making the event happen.
– We would also like to thank the supporting organizations: Sciex, Eurofins, Tohoku University, TUM, Helmholtz Munich, and Harvard University.

Dr. Matthias Scheffler
Chief Business Officer,
Chief Scientific Officer

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