Precision Omics Initiative Sweden (PROMISE) Symposium

October 28-29 2024, Uppsala

October 28th	
8:30-9:00	**Registration and coffee**
	Morning session chair: Anders Kämpe
9:00-9:45	**Tuuli Lappalainen,** KTH, SciLifeLab & NY Genome Center **Precision Omics Initiative Sweden**
9:45-10:15 *Zoom*	**Daniel MacArthur,** Director, Centre for Population Genomics, a joint initiative between the Garvan Institute of Medical Research and Murdoch Children's Research Institute **Adventures in translational genomics: lessons from the USA and Australia**
10.15-10:45	Coffee break
10:45-11:15	**Lili Milani**, Head of Estonian Biobank, Professor of Pharmacogenomics, University of Tartu **Return of results to Estonian Biobank participants: from clinical studies to an online portal**
11.15-12.15	***Building on existing precision omics projects in Sweden*** Tove Fall, Uppsala University: **Host genetic determinants of the gut microbiome composition** Tobias Sjöblom, Uppsala University: **Multi-omics analyses of colorectal cancers: biomarkers for prognosis and early** detection

	Mathias Uhlen, Royal Institute of Technology (KTH): **The creation of an open access SciLifeLab-based Human Disease Blood Atlas based on next generation proteome profiling**
12.15-13.15	Lunch
	Afternoon session chair: Åsa Johansson
13.15-13.45	**Dr. Cathryn Lewis**, Professor of Genetic Epidemiology and Statistics, King's College London **Challenges and opportunities for polygenic scores in psychiatric disorders**
13:45-14:45 **(Short talks x 3)**	**PROMISE at the cutting edge of technology** Lars Feuk, Uppsala University: **Long-read sequencing - from single samples to large cohorts and routine healthcare** Martin Enge, Karolinska Institutet: **Clonally resolved single-cell multiomics in cancer research and diagnostic** Janne Lehtiö, Karolinska Institutet: **Large scale proteome analysis from plasma to proteogenomics**
14:45-15:15	Coffee break
15:15-15.45 *Zoom*	**Erik Ingelsson,** Chief Scientific Officer, Wave Life Sciences **How genomics is transforming drug discovery and development**

15:45-17:00	***PROMISE – Key components*** Richard Rosenqvist, Karolinska Institutet: **GMS as a building block in PROMISE** Bengt Persson, Uppsala University: **European initiatives for national and international data storage and management** Anna Lindstrand, Karolinska Institutet: **Navigating Consent in Precision Healthcare** Margareta Haag, Swedish Network against Cancer: **Patient perspectives and co-creation in research and precision healthcare**
17:00-18:00	**Drinks reception**
October 29th 2024	
8:30-9:00	**Registration and coffee**
	Day 2 chair: Bo Jacobsson
9:00-10.30	***PROMISE – Targeted workshops*** **Cancer** Chair: Tobias Sjöblom, Uppsala University **Rare diseases and clinical omics** Chair: Anna Lindstrand, Karolinska Institutet **Population cohorts and complex traits** Chair: Colum Walsh, Linköping University
10:30 – 11:00	Coffee break
11:00-12:00	Workshop summary presentations and discussion
12.00-12.45	Lunch

12:45-13:15	**Samuli Ripatti,** Director, Institute for Molecular Medicine Finland FIMM, University of Helsinki, Professor of biometry, Faculty of Medicine, University of Helsinki, Affiliated Faculty Member at Massachusetts General Hospital and Broad Institute of MIT and Harvard **Large-scale biobanks in the service of translational genomic research**
13:15-13:45 *Zoom*	**Dr. Hilary Martin**, Wellcome Sanger Institute, UK **Insights into the genetic architecture of rare neurodevelopmental disorders from large cohorts**
13:45-14:15	**Dr. Kerstin Lindblad-Toh**, SciLifeLab and Department of Medical Biochemistry and Microbiology at Uppsala University as well as Vertebrate Genomics, Broad Institute of MIT and Harvard **Using evolutionary constraint to identify mutations and genes underlying complex diseases and cancer**
14:15-14:30	**Tuuli Lappalainen,** KTH, SciLifeLab & NY Genome Center **Concluding remarks**
14:30-15:00	Coffee mingle

Invited speaker bios

Daniel MacArthur is a human genomicist with over two decades of experience at the interface between human biomedicine, large-scale genomics, and data science. From 2012-2019 he served as Co-Director of the Medical and Population Genetics Program at the Broad Institute of MIT and Harvard. During this period he co-directed the Broad Institute's Centre for Mendelian Genomics, overseeing the generation and analysis of genomic data from over 10,000 individuals from rare disease families, and also led the development of the Genome Aggregation Database (gnomAD), the largest and most widely-used data set of human exome and genome sequence data, which has collated data from over 800,000 sequenced individuals. In 2020 he returned to Australia as the inaugural Director of the Centre for Population Genomics, a joint initiative between the Garvan Institute of Medical Research in Sydney and Murdoch Children's Research Institute in Melbourne. His team leads a number of national programs including OurDNA, which is working with under-represented Australian communities from the Pacific, South-East Asia, the Middle East, and Africa to build a long-term genomic resource spanning over 10,000 diverse individuals. He also leads the Australian Alliance for Secure Genomics and AI in Rare Disease (AASGARD) consortium, which works with research and clinical collaborators to test and deploy new analytical approaches for the diagnosis of severe genetic disorders.

Lili Milani is the Head of the Estonian Biobank and Professor of Pharmacogenomics at the University of Tartu. She has a PhD degree in molecular medicine from Uppsala University, Sweden. Her research focuses on the prevention and treatment of cardiovascular diseases, psychiatric conditions and how genetic variation can influence drug response, including adverse drug reactions. She is convinced that healthcare can be considerably improved by data-driven precision medicine and is actively participating in the design and implementation of personalized medicine in Estonia in close collaboration with the Ministry of Social Affairs and the Estonian Health Insurance Fund.

Cathryn Lewis is Professor of Genetic Epidemiology and Statistics at King’s College London and Head of Department at the Social, Genetic and Developmental Psychiatry Centre. She serves as Executive Director of the Psychiatric Genomics Consortium, where she co-chairs the Major Depressive Disorder Working Group. She leads the Wellcome-funded AMBER study, which aims to identify the causal mechanisms of antidepressants. With over 400 publications in statistical genetics and methodologies for complex disorders, her research focuses on characterising the genetic contributions to human health, particularly in the areas of polygenic scores, psychiatric disorders, and pharmacogenetics.

Erik Ingelsson is the Chief Scientific Officer of Wave Life Sciences (Nasdaq: WVE), a biotechnology company focused on unlocking the broad potential of RNA medicines to transform human health. Wave’s RNA medicines platform combines multiple modalities, chemistry innovation and deep insights in human genetics to deliver scientific breakthroughs that treat both rare and common disorders. Between 2000 and 2024, Dr. Ingelsson had various leadership roles at GSK, including being the Senior Vice President of Genomic Sciences, which was an organization of ~320 scientists with world-leading skills and capabilities in human

genetics, computational biology, omics technologies and gene editing, responsible for harnessing the latest methods and technologies in genomics to discover and validate novel drug targets, and to drive more successful development of the next generation of drugs. Before joining GSK, Dr. Ingelsson was Professor of Medicine at Stanford University. His lab used human genetics and functional genomics to discover novel biology and drug targets related to insulin resistance and related conditions, such as obesity, MASH and cardiovascular disease. His work combined large-scale human genetics studies with in-depth pre-clinical validation work with an emphasis on cardiometabolic diseases. Dr. Ingelsson obtained his MD and PhD at Uppsala University, Sweden. After clinical internship and residency, he did a post-doc at the Framingham Heart Study before joining the faculty at Karolinska Institutet (Stockholm, Sweden), where he was appointed Professor of Cardiovascular Epidemiology in 2010. He was also Professor of Molecular Epidemiology at Uppsala University and was a visiting professor at Oxford University. He has published over 400 peer-reviewed original articles with a h-index of 147, and have received a large number of European and US grants, awards and prizes.

Samuli Ripatti, PhD, is the Director of the Institute for Molecular Medicine Finland (FIMM), HiLIFE, Professor of Biometry at the Faculty of Medicine, University of Helsinki, and Affiliated Faculty Memberat Massachusetts General Hospital and Broad Institute of MIT and Harvard. He is chairing the Research Council Finland's Centre of Excellence in Complex Disease Genetics and the EU H2020-funded Intervene Consortium. His research group studies genetic variation in the Finnish population and its effects on common complex disease risks and management. He is one of the pioneers in developing and testing the clinical translation of polygenic risk scores in disease prevention and early detection. Dr Ripatti's research focuses on cardiometabolic diseases and cancers as models to learn about new disease mechanisms and to develop novel genome-based strategies for diagnosis, prevention and stratified treatment. He has played a central role in developing and testing the use of polygenic risk in prevention of cardiometabolic diseases and common cancers. He is also the PI of the GeneRISK Study, a pioneering longitudinal research project in studying the health impact of returning genetic and clinical risk for cardiovascular diseases to the study participants. He has published over 450 articles and book chapters.

Hilary Martin started her university studies in Australia and then completed her PhD with Peter Donnelly at the Wellcome Trust Centre for Human Genetics in Oxford in 2015. There, she worked on an eclectic set of projects ranging from rare disease genomics to population genetics of the platypus. She moved to the Sanger Institute initially as a postdoc with Jeff Barrett, and then started her own group there in 2018. Her group analyses large-scale genetic and electronic health record data to explore the genetic architecture of neurodevelopmental disorders and traits, as well as complex diseases in populations with high levels of parental relatedness (consanguinity).

Kerstin Lindblad-Toh is a professor in comparative genomics at Uppsala University and Scientific Director of Vertebrate Genomics at Broad Institute of MT and Harvard. Her work focuses on comparative genomics and comparative genetics. She co-led the Zoonomia Project which sequenced, aligned and analyzed 240 mammals to understand mammalian evolution and to identify functional positions in the human genome. This project was published in 11 papers in

Science in 2023. Dr Lindblad-Toh also works in canine and human genetics, identifying risk factors in for example amyotrophic lateral sclerosis (ALS), obsessive compulsive disorders (OCD) and multiple cancers. Evolutionary constraint is used to identify functional mutations important for disease.