Data-Driven Precision Medicine and Diagnostics Research Area Symposium

AGENDA

Time	Session/Title of the presentation	Presenter
10.00	Registration	
10:15	Welcome and Introduction to Precision Medicine and Diagnostics Research Area	Janne Lehtiö
10:30	Scientific presentations – Precision Medicine – Chair: Fulya Taylan	
	Opening Keynote: FinnGen: a public-private partnership improving human health through genetic research	Mervi Aavikko
	Genomic Medicine Sweden: Lessons learned and future possibilities within the Swedish health data space	Per Sikora
	Implementation of an academic clinical decision support system for precision oncology: the Molecular Tumor Board Portal (Digital)	David Tamborero
11:40	Lunch	
12:45	Scientific presentations – Imaging and Al for Precision Medicine -Chair: Daniel Lundqvist	
	AIDA – supporting AI research in diagnostic imaging	Claes Lundström
	Al-based precision pathology – scalable solutions for cancer patient stratification and phenotyping	Mattias Rantalainen
	Deep learning for PET image analysis	lda Häggström
13:35	Short break	
13:40	Scientific presentations – Digital Twins in Healthcare – Chair: Päivi Östling	
		Liesbet Geris
	M4-health and digital twins: bring a digital copy of yourself with you throughout your health journey	Gunnar Cedersund
	Towards precision prevention of rheumatoid arthritis; novel tools and novel results	Lars Klareskog
14:35	Coffee break	
15:00	SciLifeLab and DDLS Precision Medicine and Diagnostics -Chair: Janne Lehtiö	
	Introduction to SciLifeLab and DDLS Program	Olli Kallioniemi
	SciLifeLab's Data Center and Precision Medicine & Diagnostics Data Science Node	Johan Rung
	Bioinformatics support for Precision Medicine and Diagnostics	Pär Engström
	Q&A session	Janne Lehtiö
16:00	DDLS Fellows - Chair: Åsa Johansson	
	Deep lipidomics profiling provides potential biomarkers for precision nutrition approaches in cardiometabolic disease prevention	Clemens Wittenbecher
	Integrative multi-omics for precision medicine	Wen Zhong
	Harnessing the promise of next-generation plasma profiling for pan-cancer diagnostics	Fredrik Edfors
	Data-driven approaches toward patient-tailored treatment of neurodegenerative disease	Jacob Vogel
17:00	Closing Keynote -Chair: Janne Lehtiö	
	The need to model OMICs and routine (para)clinical data together – CLL as an example (Digital)	Carsten Utoft
17:30 - 18:30	· Networking with drinks	Niemann

Speaker's biography:



Claes Lundström's primary research focus is to develop methods that enable new levels of accuracy and efficiency within data-intensive diagnostics such as medical imaging. The efforts are concentrated at the crossroads of machine learning, visualization and human-computer interaction in demanding clinical settings.

Lundström's research is carried out within his role as Adjunct Professor at the Center for Medical Image Science and Visualization (CMIV) at Linköping University.

He is the founder and Arena Director for the Analytic Imaging Diagnostics Arena (AIDA), a national center for research and innovation in artificial intelligence based at CMIV, which started in 2017.

He is one of the leaders of the digital pathology research at CMIV, with leading roles in national and international efforts.

In parallel with his university work, Lundström holds the position as Research Director at Sectra AB.

Wen Zhong is the new DDLS fellow at Linköping University and SciLifeLab.

She received her Ph.D. degree from Chinese Academy of Sciences and joined Mathias Uhlen's lab in SciLifeLab as a postdoc in 2018. There she also played an important role in the Human Protein Atlas program.

Her research mainly focuses on the integration of multi-omics, the interplay between genetics and phenotypes, and the development of data-driven strategies/tools for precision medicine. The aim is to investigate the individualized molecular biomarkers for the estimation of disease risks, early diagnosis of disease, stratification of drug treatment response, and disease progression monitoring.

To this end, advanced artificial intelligence (AI) models for risk stratification and disease management platforms will be developed to improve the standard-of-care in the prevention and management of human diseases.





Clemens Wittenbecher is Assistant professor in precision medicine and diagnostics at Chalmers University of Technology and DDLS fellow since July 2022. He holds master's degrees in human nutrition sciences (University of Potsdam) and Epidemiology (Charité University Medicine Berlin).

His doctoral studies at the German Institute of Human Nutrition Potsdam-Rehbruecke focused on the development of graphical modeling and causal inference methods to link metabolomics networks to diet and cardio-metabolic disease risk.

Clemens postdoctoral and research associate positions at the German Institute of Human Nutrition and the Harvard School of Public Health provided advanced training in nutritional metabolomics, multi-OMICs, and precision nutrition research.

His current research leverages metabolomics and multi-omics data in prospective cohorts and dietary intervention trials to link diet to cardiometabolic disease risk with a specific focus on precision nutrition approaches.

David Tamborero is a senior scientist at the Karolinska Institutet dept. of Oncology-Pathology.

He is trained as an engineer in Electronics and engineer in Telecommunications, holds a Master degree in Bioinformatics and received a PhD in Medicine.

His current research is focused on the molecular mechanisms of tumor progression and drug response, and he has extensive experience in developing tools to support the prospective clinical decision making in different routine and investigational settings.

He participated as co-author in more than 60 peer-reviewed manuscripts (h-index of 45), including publications in top-tier journals that accumulates more than 13.000 citations.





Fredrik Edfors graduated from the Royal Institute of Technology with a Master of Science and Engineering and thereafter with a Ph.D. in Biotechnology.

He joined Professor Michael Snyder's lab at the Stanford University School of Medicine where he worked on strategies to better understand metabolic disorders and obesity.

He worked on cutting-edge at-home sample techniques paired with longitudinal personal omics profiling.

The Edfors lab is primarily engaged with blood-based protein profiling and the data-driven integration of extensive data resources that have been produced both internally as well as externally, including various analytical platforms and national registries.

The Edfors lab is now involved in analyzing the deep plasma profiles of patients diagnosed with various cancers using PEA and mass spectrometry-based strategies.

Gunnar Cedersund heads a cross-disciplinary research group at the Department of Biomedical Engineering (IMT) at Linköping University. The heart of this group (15+ people) does hybrid mathematical modelling, combining machine learning with mechanistic small- and large-scale models. These models are developed using both pre-clinical and experimental data of various types, which are collected both by others within the same group, and by numerous collaborators. These models are made available for preventive and patient-centric care, as well as for drug development and medical pedagogics, using innovative eHealth technologies.

For the last 20 years, Cedersund has developed mechanistic mathematical models for all of the main organs in the human body: heart, liver, fat, brain, etc. Lately, these models have combined into an interconnected model for the body as a whole. This interconnected model can be made specifically for each individual, and is then called a digital twin. This digital twin technology employs a hybrid approach, which combines the mechanistic simulation models with machine learning and bioinformatics models. This allows a patient, doctor, or other end-user to look inside the body of a patient, as it is now, ranging from the whole-body to the intracellular level. This also allows for simulations of different future scenarios, ranging from ms to years, and can simulate e.g. the risk of a stroke, depending on choice of diets, exercise, and certain medications. The models are thus of an M4-nature: multi-level, multi-timescale, mechanistic, and multi-organ. This digital technology is generalizable to future new usages of data and application areas, and thus goes beyond traditional narrow Al, to general, explainable Al. The digital twins are made available for end-users via a backend, which can be connected to a series of different eHealth apps. In this way, the same digital twin will be able to follow a patient across their health journey: from normal day activities like learning, concerts, and exercise, to preventive care, treatment planning, and rehabilitation.





I am an Associate Senior Lecturer and SciLifeLab DDLS fellow at Lund University. The focus of my research up to this point has been on using neuroimaging and multi-omics data to better characterize the onset and trajectory of neurodegenerative diseases. This research has been conducted with an emphasis on PET imaging, which I was trained to process and analyze at the University of California, Berkeley. I completed my PhD at the McGill Centre for Integrative Neuroscience in Montreal, supported by a Vanier Doctoral Scholarship, where my attention shifted to using machine learning, imaging-transcriptomics and big data neuroinformatics to synthesize insight from multimodal data. I completed a postdoctoral fellowship at the Lifespan Informatics & Neuroimaging Center at the University of Pennsylvania, where I began looking for traces of neurodegenative disease risk and resilience across the human lifespan. In my analyses, I like to apply unsupervised algorithms that "let the data speak", and common themes of my research involve parsing disease heterogeneity and tracking early brain changes in response to pathology. My lab at Lund U opened in December of 2020, and will focus on implementing statistical learning paradigms in order to discover early disease processes, to understand individual differences in disease expression, and to predict clinical outcomes.

Lars Klareskog is senior professor of rheumatology at Karolinska Institutet (KI) and has been the chairman of the academic department of rheumatology at KI between 1993 and 2012 and been chairman of Medicine at Karolinska Institutet and Director of the center for Molecular Medicine at Karolinska. His main research interests are in translational medicine, i.e. in understanding etiology and molecular pathogenesis of inflammatory rheumatic diseases and to use such knowledge for the development of therapy and prevention of these diseases. More specifically, he has for many years investigated how genes and environment/lifestyle interact in the etiology and molecular pathogenesis of RA and found that airway exposures including smoking can trigger autoimmunity, in particular antibodies against citrullinated proteins (ACPA).

These studies have laid the ground for an etiological hypothesis for RA, where a mucosal origin of pathogenic autoimmunity is central. Data from these studies, together with many other observations on the emergence of autoimmunity prior to the development of clinical arthritis have enabled efforts towards prevention of the disease in individuals with symptoms and autoimmunity that predict a high risk for development of RA.





Since 2013, Mattias Rantalainen leads the Predictive Medicine group at the Department of Medical Epidemiology and Biostatistics at Karolinska Institute in Sweden.

Dr. Rantalainen completed his PhD at Imperial College London with a thesis focused on multivariate pattern recognition methods and their applications in the biomedical domain, followed by postdoctoral research at the Department of Statistics and at the Wellcome Trust Centre for Human Genetics at University of Oxford.Dr. Rantalainen focus on medical research that is driven by statistical machine learning, artificial intelligence (AI) and large population representative data sets.

His research is centered on projects in areas of cancer precision medicine and computational pathology. Dr. Rantalainen leads the CHIME project at Karolinska Institute, focused on large population representative studies in computational pathology. He is the coordinator of the Swedish AI Precision Pathology (SwAIPP) consortium, and the ABCAP consortium, which are both focused on developing and translating AI-based computational cancer precision pathology.



Carsten Utoft Niemann is a hematologist, chief physician and associate professor at Copenhagen University with +10 years hands-on experience in hematology research laboratories across Europe and US.

Chairing the Nordic CLL Study Group and heading the CLL research laboratory and clinical research program at Rigshospitalet, Copenhagen, Denmark. Combining genetic and functional characterization of CLL and microenvironmental cells, MRD testing and diagnostic work up for CLL based on the EuroMRD and ERIC collaborations, along with epidemiological studies. Impacting current practice in CLL, co-authoring ESMO guidelines, TP53 guidelines, flowcytometry- and MRD-guidelines for CLL in addition to 100 peer-reviewed publications.

Founding member for the clinical GAIA/CLL13, VISION/HO141, HO158, HO159 and CLL17 trials, thus leading the way for testing targeted therapy in CLL. Founder of the PreVent-ACaLL phase 2-3 trial, the first Machine Learning based clinical trial in CLL. Thus, applying a uniquely strong background across all the disciplines needed for translational research in CLL feeding into extended decision matrices for lymphoid malignancies based on medical Artificial Intelligence for precision medicine.

Ida Häggström is an Associate Professor in the Computer Vision group at the department of Electrical Engineering at Chalmers University of Technology, working with machine and deep learning techniques for medical image analysis.

She completed two Master's degrees in Engineering Physics followed by Medical Physics, and proceeded with a PhD in Medical Physics at Umeå University, graduating in 2015. She then moved to Memorial Sloan Kettering Cancer Center in New York, USA for a postdoctoral fellowship, followed by working as a Research Associate and Senior Research Scientist, and returned to Sweden and Chalmers University of Technology in late 2021.

Ida collaborates closely with clinical researchers on projects to diagnose, predict and prognosticate different diseases, mainly cancer. She works primarily with images from positron emission tomography (PET), but also computed tomography (CT) and magnetic resonance imaging (MRI), with a focus on devising new and better problem-solving methods for challenging computer vision tasks in medical imaging.





Mervi Aavikko, PhD, Adjunct professor (molecular genetics) has over a decade of experience in translational cancer research. After receiving her PhD, about molecular mechanisms of inherited predisposition to tumors, in 2014 from the University of Helsinki, she worked at Novartis Finland, as a Medical Science Liaison in Oncology, and patient association advocate.

Passion for human genetics drove her back to research, and during 2017-2020 she worked as a post-doctoral researcher in the Tumor Genomics Group, University of Helsinki, concentrating on the genetics of intestinal cancers.

Mervi Aavikko has published over 30 peer-reviewed articles and she has received many personal grants. She has supervised several students and currently is a co-supervisor of two PhD candidates. Since November 2019 she has worked as the project manager of the FinnGen research project, combining genome and digital healthcare data on eventually 500,000 Finns. Her main tasks in FinnGen concern management of the co-operation of FinnGen teams and the science related administrative matters.



Per Sikora is head of Bioinformatics and Data Centre at the Sahlgrenska Academy, Head 1st engineer at the Sahlgrenska University Hospital, Clinical Genetics and Genomics and Chair of Informatics and Infrastructure and part of the management group in Genomic Medicine Sweden.

Expert group and adjunct members



Janne Lehtiö (chair and member of the DDLS steering group)

Affiliations: SciLifeLab / Department of Oncology-Pathology, Karolinska Institutet

Keywords: Cancer proteogenomics, precision medicine, DDLS steering group member

Gunnar Cedersund

Affiliations: Department of Biomedical Engineering (IMT), Linköping University

Keywords: Digital twins, innovative eHealth technologies

Sven Nelander

Affiliations: Department of Immunology, Genetics and Pathology, Neuro-Oncology, Uppsala University

Keywords: Data-driven cancer research, large-scale computation

Patrik Georgii-Hemming

Affiliations: Karolinska University Hospital

Keywords: Medical informatics, healthcare data

Lars Klareskog

Affiliations: Rheumatology Unit, Department of Medicine, Karolinska Institutet / Karolinska University Hospital

Keywords: Prevention, digitilisation, EIT Health

Johan Trygg

Affiliations: Department of Chemistry, Umeå University, Umeå, Sweden / Corporate Research, Sartorius, Umeå

Keywords: DDLS UmU Reference group member, DDLS PM Research Area member, DDLS DSN&WABI UmU Lead

Francis Lee

Affiliations: Technology and Social Change at the division for Science, Technology and Society, Chalmers

Keywords:WASP-HS, social and cultural aspects of digitalization and knowledge production

David Gisselsson Nord

Affiliations: Division of CG, LU / Section for pathology, Lund, Medical Services, Region Skåne, Childhood Cancer Working Group, GMS

Keywords: DDLS fellow recruitment, clinical applications, Genomic Medicine Sweden

Päivi Östling

Affiliations: SciLifeLab / Dept of Oncology-Pathology, Karolinska Institutet

Keywords: Precision Medicine, Cancer, Precision Medicine Capability















