SciLifeLab

Clinical Genomics 10th Anniversary Symposium

April 10 – April 11, 2025
Conference Centre Wallenberg, Gothenburg

“Genomic Technologies Transforming Precision Diagnostics”

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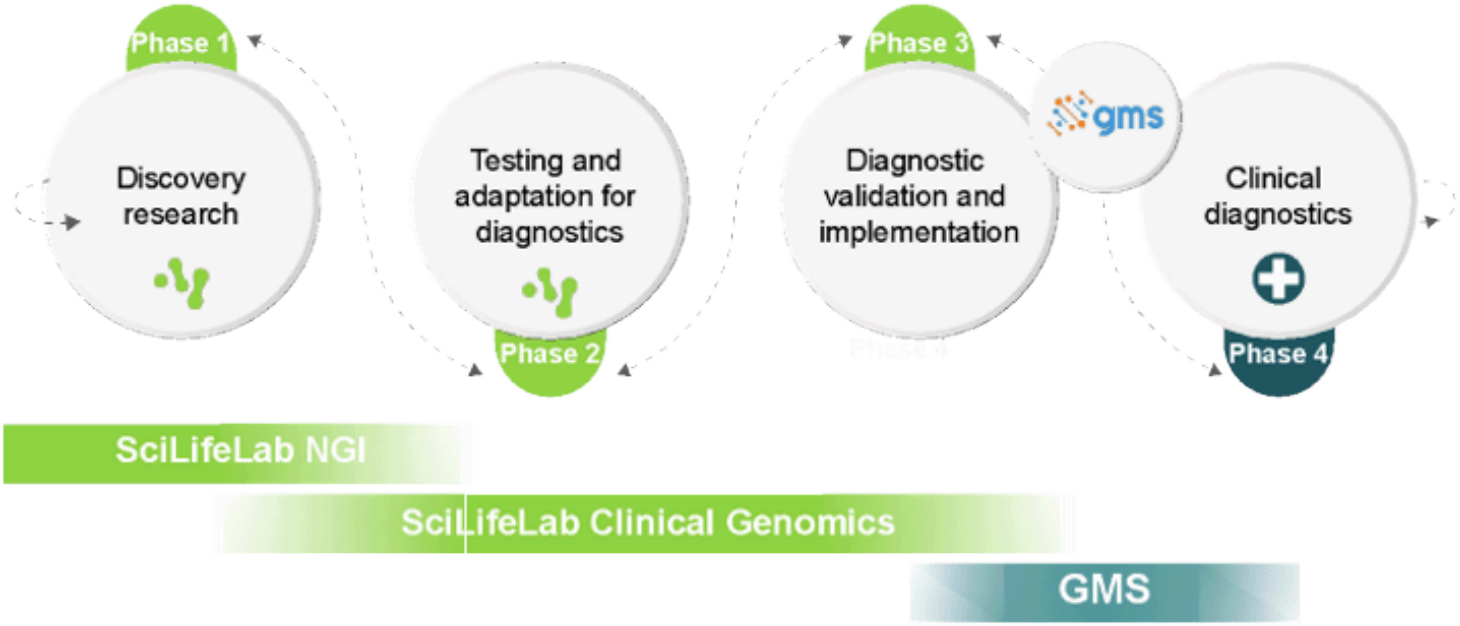
Transforming Precision Diagnostics

The national Clinical Genomics (CG) platform was established to facilitate the translation of new genomic techniques, such as next-generation sequencing (NGS), into clinical use. The overall aim of the platform is to meet the needs from translational researchers and healthcare by providing service and expertise to promote and adapt the use of genomic techniques in routine diagnostics.

Bridging Translational Research and Routine Diagnostics



The CG platform operates in phase 2 and 3 and provides expertise, technology development, and services to translational and clinical researchers, as well as clinical trials. The CG Platform also works in close collaboration with National Genomics Infrastructure (NGI) in multiple diagnostics development projects and with Genomic Medicine Sweden (GMS), the national initiative for precision medicine, to facilitate the implementation of genomics-based precision diagnostics and precision medicine in Sweden.

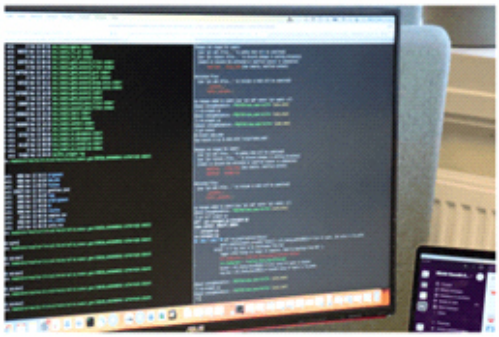
Harnessing genomic technologies for Precision Medicine

Genomic technologies hold promise in clinical diagnostics for cancer, inherited, and communicable diseases, generating vast patient data. Yet, challenges include complex workflows, specialized equipment, and extensive data analysis. Clinical Genomics integrates expertise for secure data management, processing, and accurate test interpretation, aligning with healthcare standards.

Fostering National Competence for Healthcare Innovation

A primary goal of the platform is to function as a national competence center for genomics analyses, while collaborating with healthcare institutions nationwide. Clinical Genomics aims to disseminate current information on updated protocols, guidelines and ethical standards for interpreting test results using these advanced technologies.





National outreach with nodes at all seven Swedish medical faculties

Given that Swedish healthcare is organized into autonomous regions, the national coverage is critical for ensuring equal access to services provided by the platform and accelerating the implementation of new diagnostics in healthcare throughout the country. Each node works in close collaboration with the respective university hospital.

Technology development

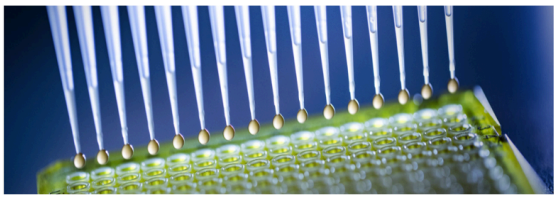
We drive our technology development through eight translational Technology Focus Areas ensuring coordination within the platform as well as long-term competitive service offering to researchers, clinical trials, and healthcare at the international forefront in the specific technology domains.

Data analytics and AI

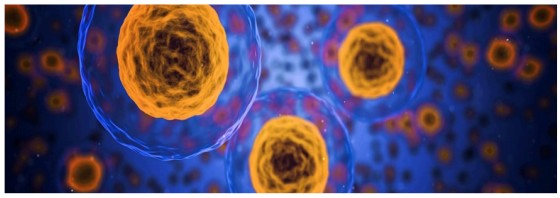
Long-read sequencing

Multi-modal omics

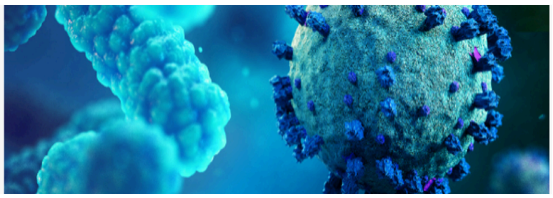
Epigenomics



Ultrasensitive Detection



Single-cell omics



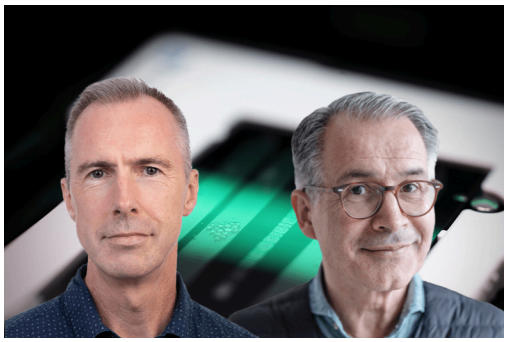
Metagenomics

Spatial omics

Celebrating a decade of impact: SciLifeLab's Clinical Genomics platform turns 10

SciLifeLab's Clinical Genomics platform is marking its 10-year anniversary, celebrating a decade of innovation, collaboration, and impact on research and healthcare. With nodes established at all seven of Sweden's medical faculties, the platform today plays an important role in providing expertise and service to clinical researchers in genomics-based assays. In addition, Clinical genomics initiated Genomic Medicine Sweden (GMS), was vital in the pandemic response efforts and has today evolved into the cornerstone of precision diagnostics in Sweden.

Read the full news piece with reflections from outgoing director Thoas Fioretos and incoming director Colum Walsh here: https://www.scilifelab.se/news/celebrating-a-decade-of-impact-scilifelabs-clinical-genomics-platform-turns-10/



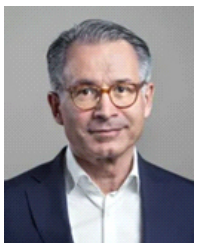
Contact

E-mail:
info-clinical-genomics@scilifelab.se

Webpage:
https://clinical-genomics.scilifelab.se

Organizing Committee

Thoas Fioretos
Platform Director, Clinical Genomics (until Dec 31, 2024); Lund University



Lucia Cavelier
Platform Co-Director, Clinical Genomics (until Dec 31, 2024); Uppsala University

Colum Walsh
Platform Director, Clinical Genomics (from Jan 1, 2025); Linköping University

Malin Melin
Platform Co-Director, Clinical Genomics (from Jan 1, 2025); Uppsala University

Marcela Davila
Platform Coordination Officer, Clinical Genomics; University of Gothenburg

Eva Berglund
Platform Strategy and External Relations Officer, Clinical Genomics; Uppsala University

Maria Smedh
Gothenburg Site Coordinator; University of Gothenburg

Program - April 10

8:30-10:35	**Introduction Session**
8:30-9:00	Coffee and Registration
9:00-9:05	Welcome address Thoas Fioretos, outgoing Platform Director Clinical Genomics
9:05-9:35	10 years with Clinical Genomics: Changing the landscape of genome-based precision diagnostics Valtteri Wirta, Platform Scientific Director, Clinical Genomics Stockholm
9:35-10:05	SciLifeLab: Enabling Scientific Discovery Through Cutting-Edge Infrastructure Annika Jenmalm Jensen, Infrastructure Director SciLifeLab
10:05-10:35	My diagnostic odyssey: Why is a correct diagnosis important for the patient? Helene Cederroth, Founder and Chair Wilhelm Foundation
10:35-11:30	**Coffee break**

Program - April 10, cont.

11:30-12:40 **Session 1: Current and emerging next-generation sequencing technologies**
Chairs: Anna Lyander, CG Stockholm, and Malin Melin, CG Uppsala

11:30-12:00 Keynote: Sequencing technologies now and in the future
Lars Feuk, Platform Co-Director, National Genomics Infrastructure, Uppsala

12:00-12:20 Nucleic acid analysis in cancer management using liquid biopsies
Anders Ståhlberg, Professor, University of Gothenburg

12:20-12:40 Rapid diagnosis of leukemic aberrations using nanopore sequencing
Tatjana Pandzic, Clinical Laboratory Geneticist, Uppsala University

12:20-14:00 **Lunch**

14:00-15:10 **Session 2: Genomics for pathogen detection and pandemic preparedness**
Chairs: Paula Mölling, CG Örebro, and Valtteri Wirta, CG Stockholm

14:00-14:30 Keynote: With a mission to fight a pandemic outbreak: Lessons learned?
Lars Engstrand, Professor, Karolinska Institutet, Stockholm

14:30-14:50 Nationwide multicentre study evaluating Nanopore sequencing for bacterial 16S rRNA-species identification
Sofia Brunet, PhD student, Sahlgrenska University Hospital, Gothenburg

14:50-15:10 Integrative Outbreak Simulation: A One Health Approach for Enhanced Pandemic Preparedness
René Kaden, Associate Professor, Akademiska sjukhuset, Uppsala

Program - April 10, cont

15:10-16:00 **Coffee break**

16:00-16:30 **Keynote**
Chair: Colum Walsh, CG Linköping
Genomic Medicine Sweden: Impacting Swedish Precision Medicine
Richard Rosenquist, Director Genomic Medicine Sweden (online)

16:30-17:30 **Session 3: Advancing Genomics: Insights from Industry Leaders**
Chairs: Ka-Wei Tang, CG Gothenburg, and Linda Köhn, CG Umeå

16:30-16:45 Roche Sequencing Solutions - Introducing Sequencing by Expansion
Joel Jonasson, Commercial Specialist, Sequencing Roche Diagnostics Scandinavia AB

16:45-17:00 Take the leap into multiomics with Illumina innovation
Filip Stern, Executive Sales Specialist, Illumina – North

17:00 -17:15 What You're Missing Matters: Catching the Unnoticed with Nanopore Sequencing
Christos Coucoravas, Field Application Scientist, Oxford Nanopore Technologies, Nordics

17:15 - 17:30 Unlocking new insights in oncology research with AMP technology
Moa Persson, NGS Clinical Sales Specialist, Integrated DNA technologies, Archer

17:30 Welcome drink followed by dinner

Program - April 11

9:00-9:30 **Keynote**
Chair: Lucia Cavelier, outgoing co-Platform Director Clinical Genomics
Genomics technologies that will shape the future of precision diagnostics
Joakim Lundeberg, Professor, KTH Royal Institute of Technology, Stockholm

9:30-10:40 **Session 4: Translational Single cell and Spatial Omics**
Chairs: Anna Hagström, CG Lund, and Julia Bräunig, CG Lund

9:30-10:00 Keynote: What can we learn from the human cell atlas?
Olli Dufva, Postdoctoral fellow, Wellcome Sanger Institute, UK

10:00-10:20 Mapping ex vivo drug responses in single cells
Jessica Nordlund, Associate Professor, Uppsala University

10:20-10:40 Molecular understanding of response to immune checkpoint blockade in melanoma
Göran Jönsson, Professor, Lund University

10:40-11:20 Coffee

11:20-12:30 **Session 5: Diagnostic Analytics and Informatics**
Chairs: Lars Palmqvist, CG Gothenburg, and Marcela Dávila, CG Gothenburg

11:20 -11:50 Keynote: Dynamic digital twins for personalised early diagnostics and therapeutics
Mikael Bensson, Principal Researcher, Karolinska Institutet. Stockholm

11:50-12:10 Digital biology: Large-scale integration of biomedical data to decipher biology and new treatments
Araz Rawshani, Associate Professor, University of Gothenburg

12:10-12:30 AI in Clinical Bioinformatics
Marcin Kierzcak, NBIS expert, Uppsala University

12:30-14:00 **Lunch**

Program - April 11, cont.

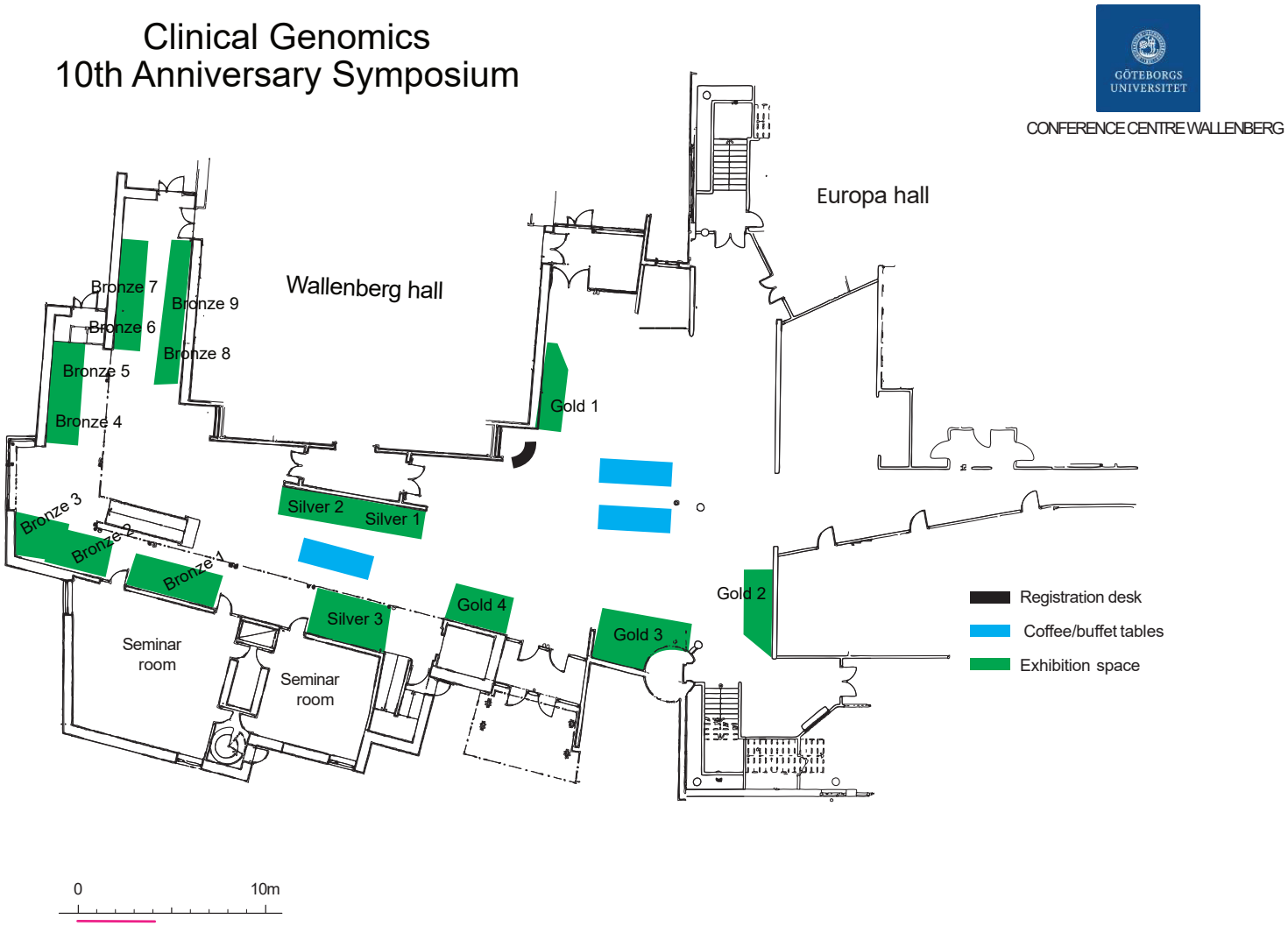
14:00-14:30 **Keynote**
Chairs: Lucia Cavelier, outgoing co-Platform Director CG and Thoas Fioretos, outgoing Platform Director CG
Implementing genomic medicine at a national level
Matt Brown, CSO Genomics England; UK (online)

14.30-15.00 **Panel discussion**
Clinical Genomics: Challenges, opportunities, and the next decade
Mikael Benson, Lucia Cavelier, Helene Cederroth, Lars Feuk and Thoas Fioretos
Moderator: Colum Walsh

15:00-15:10 Concluding remarks
Colum Walsh, incoming Platform Director Clinical Genomics

15:10 Coffee and end of conference

Exhibitions



Gold Exhibitors

Integrated DNA Technologies	Gold 1
Oxford Nanopore Technologies	Gold 2
Illumina	Gold 3
Roche Diagnostics Scandinavia	Gold 4

Silver Exhibitors

Parse BioSciences	Silver 1
Techtum Lab	Silver 2
Triolab	Silver 3

Bronze Exhibitors

10x Genomics	Bronze 1	Proteintech Europe	Bronze 6
Thermo Fisher Scientific	Bronze 2	Bruker Spatial Biology	Bronze 7
Nordic Biolabs AB and Promega	Bronze 3	Saveen & Werner	Bronze 8
BioNordika and New England Biolabs	Bronze 4	Hamilton Nordic AB	Bronze 9
Takara Bio	Bronze 5		

Gold Exhibitors

illumina®

Roche

IDT™
INTEGRATED DNA TECHNOLOGIES

Oxford Nanopore Technologies

Silver Exhibitors

techtum

TRIOLAB

PARSE BIOSCIENCES

Bronze Exhibitors

nordic biolabs

BioNordika

SAVEEN WERNER

Promega

NEW ENGLAND Biolabs

10x GENOMICS

proteintech®

ThermoFisher SCIENTIFIC

BRUKER

TaKaRa

HAMILTON

Mikael Benson

Mikael Benson is a principal researcher at Karolinska Institutet in Stockholm, Sweden, leading the Medical Digital Twin Research Group. The clinical aims are early personalised diagnosis and treatment based on dynamic Digital Twin (DTs) of individual's disease processes. The DTs are computational models constructed by integration of clinical routine and multi-omics data down to the spatial single cell level (web sdtc.se). His interdisciplinary research spans systems medicine, computational biology, and bioinformatics, contributing to over 100 publications with significant citations. In a notable study published in Genome Medicine 2024, his team showed that DTs are ready for clinical trials of personalised treatment based on computational treatment of the DTs with thousands of drugs. Benson also co-founded AB Mavatar, a company advancing digital twin technology in medicine, with his work bridging the gap between complex disease mechanisms and modern healthcare.

Dynamic digital twins for personalised early diagnostics and

A key healthcare problem is that many patients do not respond to treatment. This may depend on the enormous complexity and heterogeneity of diseases, each of which may involve thousands of genes across multiple cell types. Those genes can vary between two patients with the same diagnosis, as well as between the same patient at different time points. Ideally, diagnostics and therapeutics should be based on characterisation of individual patients' disease processes over time. Dynamic Digital Twins may contribute by computational integration of multi-omics data down to the spatial single cell level, at different time points of a disease process.

Matt Brown

Dr. Matt Brown is a clinician-scientist who trained in medicine and rheumatology in Sydney, Australia, before completing a Doctorate of Medicine at the University of Oxford, focusing on the genetics of ankylosing spondylitis. He is a Professor of Medicine at King's College London and since 2021 has served as the Chief Scientific Officer at Genomics England.

Implementing genomic medicine at a national level

Next-generation sequencing has transformed rare disease diagnostics, significantly shortening the diagnostic odyssey. However, whole genome sequencing (WGS) provides answers for only ~30% of patients referred for genetic testing. To bridge this gap, Genomics England is exploring emerging technologies, including long-read and epigenetic sequencing, transcriptomics, proteomics, and metabolomics. These approaches aim to enhance diagnostic yield and improve patient outcomes.

Sofia Brunet

Sofia Brunet is a PhD student at Sahlgrenska University Hospital, with a background in clinical microbiology. Her research focuses on the applications of Next-Generation Sequencing in microbiology. Her latest work evaluates the potential of Nanopore sequencing for bacterial identification, exploring its feasibility as a diagnostic tool. She has co-authored work on the metagenomic detection of microbial pathogens and works closely with NGS technologies in a clinical setting.

Nationwide multicentre study evaluating Nanopore sequencing for bacterial 16S rRNA-species identification

This study evaluated the clinical utility of Oxford Nanopore Technology (ONT) sequencing for bacterial identification in a nationwide multicentre study coordinated by Genomic Medicine Sweden (GMS). Seventeen of Sweden's 21 regions in collaboration with NGI, FOHM and FOI participated, analysing bacterial mock samples alongside a standardized Quality Control for Molecular Diagnostics panel. Taxonomic identification was performed using two bioinformatics solutions: the 1928 Diagnostics commercial 16S pipeline and the open-source GMS-16S pipeline. Our work demonstrates the potential of ONT implementation into clinical laboratories. Further development of the protocol will likely enhance the utility of Nanopore sequencing for 16S rRNA bacterial species identification, improving national precision diagnostics.

Helene Cederroth

Founder and President of Wilhelm Foundation, dedicated to ending the diagnostic odyssey for People Living with Undiagnosed Diseases (PLWUD) worldwide, with a special focus on Low- and Middle-Income Countries. Co-founder and board member of the Undiagnosed Diseases Network International (UDNI) and co-founder of the Undiagnosed Diseases Network Foundation (UDNF). Co-arranged 13 UDNI Conferences alongside Dr. William Gahl of the Bethesda UDP NIH and various local hosts. Pioneered the international Undiagnosed Hackathons. Organized the first Undiagnosed Hackathon together with Karolinska UDP, and the second at Radboud umc. Wilhelm Foundation is organizing the third Undiagnosed Hackathon with Mayo Clinic in Rochester, Minnesota. Helene represents the Wilhelm Foundation in the International Rare Diseases Research Consortium (IRDiRC) and collaborates with Karolinska-UDP. An international speaker on the topic of People Living with Undiagnosed Diseases (PLWUD), Helene's work is deeply personal. She tragically lost three of her four children—Wilhelm, Emma, and Hugo—to an undiagnosed disease at the ages of 16, 10, and 6. The disease was neither thought to be hereditary nor fatal, and this profound loss ignited her relentless pursuit to ensure no family has to endure the same pain

My diagnostic odyssey: Why is a correct diagnosis important for the patient

Helene Cederroth shares her personal journey—losing three of her four children to an undiagnosed condition—and how it propelled her to become a global leader for people living with undiagnosed diseases (PLWUDs). She highlights the significance of early and accurate diagnosis, cross-sector collaboration, and the impact of the Undiagnosed Hackathon, a platform she co-initiated to foster innovative solutions. Helene underscores the need to bridge healthcare gaps, particularly in low- and middle-income countries, and calls for a unified, international approach to end the diagnostic odyssey. She urges each of us to champion hope, progress, and collective responsibility in improving outcomes for families with undiagnosed diseases worldwide.

Olli Dufva

Dr. Olli Dufva is a physician-scientist and postdoctoral fellow at the Wellcome Sanger Institute and the Cambridge Stem Cell Institute at the University of Cambridge, working in the laboratories of Prof Sarah Teichmann and Dr Mathew Garnett. He earned his medical degree and Ph.D. from the University of Helsinki, where he studied immune interactions and precision medicine in blood cancers. His research focuses on mapping immune cells across tissues, understanding genomic determinants of anti-cancer immunity, and development of improved cellular immunotherapies using single-cell genomics, organoid models, and genetic engineering. With expertise in computational analysis and functional genomics, Dr. Dufva aims to advance immunotherapy strategies for cancer treatment.

What can we learn from the human cell atlas?

The Human Cell Atlas (HCA) is a global initiative aiming to create a comprehensive reference map of all human cells as a basis for both understanding human health and diagnosing, monitoring and treating disease. The HCA's approach is providing unprecedented understanding of human cells and tissue architecture using single-cell and spatial genomics. Dr. Dufva will give an overview of recent advances on mapping cells across tissues and discuss ongoing work on integrating disease models with tissue atlases and investigating blood cell states across thousands of individuals using a population-scale single-cell

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Lars Engstrand

Dr. Lars Engstrand is a professor and senior physician at Karolinska Institutet's Department of Microbiology, Tumor, and Cell Biology. He directs the Centre for Translational Microbiome Research (CTMR), focusing on the role of microorganisms in gastrointestinal diseases and reproductive health. In 2020, he established the National Pandemic Centre at Karolinska Institutet to enhance pandemic preparedness. Dr. Engstrand's research includes the pathophysiology, diagnosis, and treatment of gastrointestinal infections, with a particular emphasis on Helicobacter pylori. He has published over 300 scientific articles and book chapters, contributing significantly to microbiome research and its implications for human health.

With a mission to fight a pandemic outbreak: Lessons learned

In March 2020 Sweden was hit by Covid-19 and the global consequence of the outbreak was unknown at that time. With substantial support from external funding agencies, our research lab was turned into a large-scale, high-throughput diagnostic and surveillance lab for almost 2,5 years. Many lessons were learned and hopefully many of them will be taken into consideration next time.

Lars Feuk

Dr. Lars Feuk is a Professor in the Department of Immunology, Genetics, and Pathology at Uppsala University, where he leads a research group focusing on genomics and neurobiology. His work centers on human genome variation, particularly the role of structural genetic variations in neurodevelopmental disorders. As Co-Director of the SciLifeLab National Genomics Infrastructure, Dr. Feuk is involved in applying advanced sequencing technologies to enhance understanding of human genome architecture and improve clinical diagnostics. He has contributed to studies utilizing long-read sequencing to resolve complex chromosomal rearrangements, advancing the field of precision medicine.

Sequencing technologies now and in the future

Over the last ten years the field of human genome sequencing has developed rapidly, with significant improvements in accuracy, throughput and decreased cost. While early development and clinical diagnostic implementation have relied almost exclusively on a single sequencing technology, the field is now going through a rapid transformation, with several new players on the market. The new and emerging technologies use different strategies to challenge the traditional sequencing market. In addition to accuracy, throughput and cost, new instruments show improved capabilities in specific aspects of sequencing including flexibility, rapid sequencing, read-length, decoding epigenetic modifications and multi-omic capabilities. The presentation will outline the current trends in the sequencing field and highlight some of the latest and emerging technologies that are challenging the current landscape of diagnostic sequencing.

Annika Jenmalm Jensen

Dr. Annika Jenmalm Jensen is the Infrastructure Director at SciLifeLab and a researcher at Karolinska Institutet. She earned her Ph.D. in Medicinal Chemistry from Uppsala University in 1998 and subsequently worked in preclinical drug discovery at Pharmacia and Biovitrum. In 2009, she became the Director of the Chemical Biology Consortium Sweden (CBCS), a position she held until 2016. Since then, she has been leading efforts to maintain and advance SciLifeLab's infrastructure, focusing on integrating cutting-edge technologies into life sciences research. Additionally, Dr. Jenmalm Jensen serves as the Deputy Chair of the MAX IV Board, contributing her expertise to the development of national research infrastructures.

SciLifeLab: Enabling Scientific Discovery Through Cutting-Edge Infrastructure

National research infrastructures play a crucial role in enabling scientific discoveries that would otherwise be possible. SciLifeLab provides access to cutting-edge technologies, expertise, and large-scale data resources, empowering researchers across disciplines. As science becomes increasingly data-driven, the future of national infrastructures will depend on seamless integration of AI, advanced analytics, and improved data sharing. This talk will highlight how SciLifeLab fosters innovation through interdisciplinary collaboration, strategic investments, and a forward-looking approach to infrastructure development. By evolving towards a more data- and AI-centric model, national infrastructures can accelerate breakthroughs in life sciences and beyond, shaping the future of research.

Göran Jönsson

Dr. Göran Jönsson is a professor of molecular oncology at Lund University, specializing in melanoma research. His work focuses on the genetic and molecular landscape of melanoma, with significant contributions to understanding how tertiary lymphoid structures—specific clusters of B cells within tumors—affect patient prognosis and response to immunotherapy. In recognition of his pioneering research, Dr. Jönsson received the Göran Gustafsson Prize in Medicine in 2022 and was awarded Cancer Research of the year from the Swedish Cancer Society in 2023 . He has also been awarded substantial grants, including SEK 31 million from the Knut and Alice Wallenberg Foundation, to further his studies aimed at enhancing the effectiveness of immunotherapy for cancer patients.

Molecular understanding of response to immune checkpoint blockade

Over the past decade, immunotherapy has become a cornerstone in enhancing survival outcomes for cancer patients. Recent studies, including our own, have provided compelling evidence that tertiary lymphoid structures (TLS) are pivotal in orchestrating anti-tumor immune responses in melanoma. These specialized immune niches are critical determinants of immunotherapy efficacy, influencing clinical outcomes in profound ways. We are now further elucidating the role, heterogeneity and cellular composition of such structures with the aim to develop new effective immunotherapies.

Rene Kaden

Dr. René Kaden is an Associate Professor at Uppsala University's Department of Medical Sciences, specializing in Clinical Microbiology. His research focuses on the epidemiology, taxonomy, and evolution of microorganisms. He coordinates the Infectious Diseases work package within Genomic Medicine Sweden for Region Uppsala and leads the Microbiology work package at Clinical Genomics Uppsala. Dr. Kaden's work includes high-throughput long-read sequencing and bioinformatics analysis to enhance the understanding of microbial genetics and improve diagnostic methods.

Integrative Outbreak Simulation: A One Health Approach for Enhanced Pandemic

The COVID-19 pandemic provided valuable knowledge, especially through collaborative projects such as the Genomic Pandemic Preparedness Portfolio (G3P). However, knowledge and skills can fade without regular training and testing. In collaboration with six Pandemic Laboratory Preparedness (PLP) partners, the Swedish Defence Research Agency (FOI), and the National Veterinary Institute (SVA), we simulated an outbreak with real clinical samples and environmental cases, applying the One Health approach. We assessed response times, diagnostics, sequencing, and scale up capacities. Progress was documented on the Swedish Pathogens Portal, identifying strengths and areas for improvement in pandemic preparedness.

Marcin Kierczak

Dr. Marcin Kierczak is a bioinformatics data scientist at the National Bioinformatics Infrastructure Sweden (NBIS) and an associate professor in bioinformatics at Uppsala University. He earned his Ph.D. in bioinformatics from Uppsala University in 2010 and joined NBIS in 2016 after postdoctoral positions at the Swedish University of Agricultural Sciences and Uppsala University. His research focuses on applying machine learning and artificial intelligence methods to genetics and population genetics, including metagenomics and ancient DNA studies. Dr. Kierczak is also involved in teaching programming, particularly in R, and co-organizes the RaukR Advanced R Programming for Bioinformatics Summer School. Additionally, he serves as the Industry Outreach Coordinator at NBIS, facilitating collaborations between academia and industry.

AI in Clinical Bioinformatics

Transforming Healthcare through Data Intelligence Marcin Kierczak, NBIS In my talk, I will provide an overview of the landscape of artificial intelligence (AI) applications in clinical bioinformatics, with a particular focus on clinical genomics. I will begin with a general introduction to AI, distinguishing it from related concepts such as machine learning (ML) and deep learning (DL). Following this, I will discuss key domain-specific features and challenges that AI scientists face in clinical settings, including model validation, explainability, interpretability, and legal and ethical considerations. After this foundational discussion, I will present an overview of various AI applications in clinical bioinformatics, emphasizing tools tailored for clinical genomics. Finally, I will showcase some relevant Swedish projects that aim to apply AI methodologies to clinical data.

Joakim Lundeberg

Dr. Joakim Lundeberg is a professor of molecular biotechnology at KTH Royal Institute of Technology and a leading figure at the Science for Life Laboratory (SciLifeLab) in Stockholm. He earned his Ph.D. in Biotechnology from KTH in 1993 and, after a postdoctoral period at the Radiumhospital in Oslo, returned to KTH as a group leader. In 2000, he was appointed professor in Molecular Biotechnology at KTH. Dr. Lundeberg's research focuses on molecular technology development, particularly in spatial omics, with applications in medicine and environmental studies. He is renowned for developing Spatial Transcriptomics, a technology that enables whole-transcriptome spatially resolved RNA sequencing in human tissues. His work has significantly advanced the understanding of gene expression within tissue architecture, contributing to insights in areas such as cancer, neurology, and human development. In recognition of his contributions, he was awarded over 30 million SEK by the Swedish Research Council's distinguished professor grant in 2025 to further his research in innovative medical technologies.

Genomics technologies that will shape the future of precision diagnostics

The technologies to assemble spatial maps that describe and explain the cellular basis of disease are quickly being more used. We have developed and established the Spatial Transcriptomics technology, in which tissue imaging is merged with spatial RNA sequencing and resolved by computational means. Our technology was the first method to provide unbiased whole transcriptome analysis with spatial information from tissue; since its initial publication, it has been used in multiple biological systems in health and disease and is the most widely applied method to study gene expression in tissue. This presentation will cover the next steps toward bringing spatial omics into personalized cancer care.

Jessica Nordlund

Dr. Jessica Nordlund is a researcher at Uppsala University's Department of Medical Sciences, where she leads the Molecular Precision Medicine research group. Her work focuses on developing and applying novel molecular approaches to analyze genomes, transcriptomes, epigenomes, and proteomes of leukemia cells. By integrating various types of omics data with machine learning, her team aims to identify molecular biomarkers to improve precision in molecular diagnoses and discover new treatment approaches. Dr. Nordlund also serves as the Managing Director for the SNP&SEQ Technology Platform at the SciLifeLab National Genomics Infrastructure in Uppsala. She earned her bachelor's degree in biology from San Diego State University in 2006 and a PhD in molecular medicine from Uppsala University in 2012. In recognition of her contributions, she received the Göran Gustafsson Prize in Medical Science at Uppsala University.

Mapping ex vivo drug responses in single cells

The goal of functional precision medicine is to tailor treatments based on individual drug responses. We have developed an approach that integrates high-throughput ex vivo drug screening with transcriptomic and spatial proteomic readouts to map leukemia cell responses to cytotoxic agents in bulk and single cells. This approach enables multiplexed drug response analysis within a single experiment, capturing transcriptional and cell-surface proteome changes at single-cell resolution. By identifying drug-tolerant subpopulations and molecular resistance mechanisms, our work provides a deeper mechanistic understanding of how individual leukemic cells and cell states respond to treatments.

Tatjana Pandzic

Dr. Tatjana Pandzic is a clinical laboratory geneticist and researcher at Uppsala University Hospital's Department of Clinical Genetics. She co-leads the hematology work package at Clinical Genomics Uppsala, focusing on implementing short and long read sequencing methods as well as digital PCR assays to enhance diagnostics and monitoring of patients with hematologic malignancies. Her research includes developing comprehensive gene panels for myeloid malignancies, improving diagnostic accuracy and enabling personalized patient monitoring. Dr. Pandzic has also contributed to studies on minimal residual disease detection post-allogeneic stem cell transplantation and the identification of recurrent mutations in chronic lymphocytic leukemia.

Rapid diagnosis of leukemic aberrations using nanopore sequencing

Genomic abnormalities facilitate the diagnosis, disease classification and prognosis in leukemias and play an important role in selection of treatment strategies.Updated guidelines for the diagnosis of leukemia recommend testing for additional genes and gene rearrangements that are not currently evaluated. In addition, it is also recommended that mutation status for therapeutically actionable targets is available within 3 to 5 days, which current methods cannot deliver.The aim of this project is to develop genetic tests for both mutation detection and detection of gene rearrangements that can deliver clinical reports in a short period of time without compromising sensitivity. For this, a targeted long-read sequencing method from Oxford nanopore technologies (ONT) was selected as it enables a rapid workflow and is easy to customize by including new regions. In project 1 we have used an amplicon-based approach to assess the mutation status of FLT3, NPM1, TP53, IDH1 and IDH2. 15 amplicons were designed to cover the regions of interest in these genes. These amplicons are amplified in 3 multiplex PCR-pools and subsequently sequenced as one library.In project 2 we have used adaptive sampling from ONT to detect gene fusions at high resolution from DNA samples. Adaptive sampling is solely software-controlled and requires no wet lab-based enrichment.

Araz Rawshani

Dr. Araz Rawshani is an associate professor at the University of Gothenburg's Institute of Medicine and a resident physician in cardiology at Sahlgrenska University Hospital. His current research leverages artificial intelligence to diagnose acute coronary syndromes, automate radiological tasks in cardiology, create clinical agents for cardiovascular medicine and use language models capable of reasoning around cardiometabolic research questions. Dr. Rawshani's work emphasizes rapid translation of research findings into clinical practice, enhancing patient outcomes.

Digital Biology: Large-Scale Integration of Biomedical Data To Decipher

The exponential growth of biomedical data, coupled with advances in artificial intelligence, will accelerate and transform our understanding of human biology. AI models can integrate vast datasets—ranging from genomics and proteomics to clinical and pharmacological data—uncovering complex physiological interactions and disease mechanisms. By strategically structuring and leveraging these resources, it is possible to accelerate drug discovery, optimize treatment strategies, and enhance precision medicine. As biomedical databases continue to expand, AI-powered digital biology will be a cornerstone of future healthcare innovation, driving breakthroughs beyond our current perception.

Richard Rosenquist

Richard Rosenquist Brandell, MD, PhD, is Professor of Clinical Genetics at Karolinska Institutet, and Senior Physician at Karolinska University Hospital, Stockholm, Sweden. He is also Director of Genomic Medicine Sweden, a national infrastructure for implementing precision medicine. By utilizing cutting-edge molecular tools, his work has led to the identification of novel prognostic and predictive biomarkers in hematological malignancies, enhancing patient risk stratification and clinical decision-making, hence paving the way for implementation of precision medicine. Richard Rosenquist Brandell is a member of the Nobel Assembly at Karolinska Institutet.

Genomic Medicine Sweden: implementing precision medicine at a national level

Precision medicine is transforming healthcare by enabling personalized treatment through high-throughput sequencing and targeted therapies. While many countries have adopted national strategies, Sweden has faced challenges due to its regionally organized healthcare. To address this, key stakeholders established Genomic Medicine Sweden (GMS), a national infrastructure integrating precision diagnostic services and secure data management. GMS focuses on rare diseases, cancer, complex diseases and infectious diseases, while addressing informatics, ethicolegal aspects, health economics, and education. Examples highlighting successful implementation of precision medicine will be shared, along with the challenges, opportunities, and future directions in this field.

Anders Ståhlberg

Dr. Anders Ståhlberg is a professor in Clinical Genomics at the University of Gothenburg and a principal investigator at the Sahlgrenska Center for Cancer Research. He also leads the Translational Genomics Platform at Sahlgrenska University Hospital, focusing on developing and implementing liquid biopsy analyses into healthcare. His research aims to understand molecular mechanisms in tumor initiation and development, with a particular focus on sarcomas and cancer diagnostics. Dr. Ståhlberg has developed several strategies for liquid biopsy analysis and single-cell profiling, contributing to advancements in non-invasive cancer diagnostics and personalized treatment approaches.

Nucleic acid analysis in cancer management using liquid biopsies

The use of liquid biopsy-based biomarker analysis emerges with clinical applications such as screening of asymptomatic individuals, diagnosis, treatment prediction, prognostication, monitoring treatment efficacy and early detection of treatment resistance as well as relapse. Digital sequencing enables ultrasensitive detection of DNA, RNA molecules derived from tumor cells as well as immune cells in body fluids. Here, we present SiMSen-Seq that can be applied to different types of nucleic acids, with tumor-informed circulating tumor-DNA analysis as a key application. Data from several ongoing studies and tumor entities will be presented to demonstrate technical solutions and clinical utility.

Valtteri Wirta

Valtteri Wirta, PhD, is a leading expert in genomics and precision medicine, serving as the Platform Scientific Director at Clinical Genomics Stockholm at SciLifeLab. He plays a key role in integrating high-throughput genomic technologies into clinical diagnostics, collaborating with Swedish healthcare to advance cancer, rare disease, and infectious disease research. With a Ph.D. in Biotechnology from KTH Royal Institute of Technology, his work emphasizes the implementation of whole genome sequencing in healthcare, significantly improving diagnostic rates. His research bridges cutting-edge genomic advancements with practical clinical applications, enhancing precision medicine and patient care.

10 years with Clinical Genomics: Changing the landscape of genome-based precision

My presentation, "10 Years with Clinical Genomics: Changing the Landscape of Genome-Based Precision Diagnostics," highlights the evolution of clinical genomics over the past decade and its transformative impact on precision medicine. I will discuss key advancements in sequencing technologies, bioinformatics, data interpretation, and integration into healthcare systems, emphasizing how these innovations have improved diagnostics and patient outcomes. Case studies illustrate real-world applications, challenges, and future directions in the field. My talk will also explore implementation hurdles, underscoring the need for collaboration between clinicians, researchers, bioinformaticians, policymakers and other competences to maximize the benefits of genomic medicine in clinical practice.

Joel Jonasson
Commercial Specialist, Sequencing,
Roche Diagnostics Scandinavia

Roche Sequencing Solutions - Introducing Sequencing by Expansion

Be the first to enter an entirely new next generation sequencing space. Explore how our novel sequencing technology has been designed to push your science further and how this technology complete the sequencing portfolio of Roche Diagnostics

Filip Stern
Executive Sales Specialist,
Illumina – North

Take the leap into multiomics with Illumina innovation

Proteomics, methylation, single cell and spatial. A short introduction to new and future Illumina launches that will expand the possibilities of multiomics to gain even greater insights.

Christos Coucoravas,
Field Application Scientist,
Oxford Nanopore Technologies, Nordics

Oxford Nanopore Technologies

What You're Missing Matters: Catching the Unnoticed with Nanopore Sequencing

In clinical genomics, what goes unnoticed can be just as critical as what is detected. Conventional sequencing methods often miss structural variants, epigenetic modifications, and low-frequency variants—gaps that can impact patient diagnosis and treatment. Nanopore sequencing, with its ability to generate ultra-long reads in real time and directly detect base modifications, offers a transformative approach to capturing these elusive genomic elements.

In this talk, we explore how nanopore sequencing is reshaping precision diagnostics by enabling comprehensive variant detection across complex regions of the genome. We will highlight its applications in rare disease, cancer, and methylation, where missing key genetic information can have a large impact in both quick turnaround time for patient results but also treatment approaches.

We will also discuss how real-time, adaptive sequencing can enhance diagnostic workflows, allowing dynamic enrichment of relevant variants without the need for extensive bioinformatic preprocessing. By leveraging the latest advances in nanopore technology, we can bridge the gaps left by conventional methods, providing a more complete genomic picture, and driving better clinical outcomes.

Join us as we showcase why what you're missing matters—and how nanopore sequencing is making the invisible, visible.

Moa Persson
NGS Clinical Sales Specialist,
Integrated DNA technologies, Archer

Unlocking new insights in oncology research with AMP technology

Integrated DNA Technologies' (IDT) Archer™ portfolio offers blood cancer and solid tumor research solutions for NGS library preparation and data analysis. AMP™ technology, the power behind IDT's Archer NGS research assays, can be used for applications in DNA, RNA, and ctDNA sequencing from a wide range of inputs, including FFPE. Our chemistry is combined with our unique and purpose-built data analysis solutions, allowing users to accurately identify both simple and complex genetic mutations--including our newly launched HRD assessment capabilities. This combination of chemistry and software analysis allows IDT to offer integrated solutions that solve common workflow and detection challenges commonly experienced with NGS approaches.

Additionally, because the primers in Archer NGS research assays function independently, panel content can be efficiently customized based on individual workflow needs—without influencing panel performance. This provides labs with targeted NGS panels that can grow as new biomarkers and panel content is identified, effectively future proofing workflows.

In this presentation, learn about the utility of Archer integrated NGS solutions (AMP chemistry and Archer Analysis) for biomarker identification, even with poor quality samples, and explore new additions to the Archer NGS portfolio including flexible comprehensive genomic profiling (CGP) and modular HRD assessment solutions.

Notes

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SciLifeLab

Clinical Genomics Platform