NSHG-PM 2023 WORKSHOP

PRECISION MEDICINE: THE BRIDGE FROM RESEARCH TO HEALTHCARE

19 - 21 JUNE 2023 · TALLINN · ESTONIA









THANKS FOR SUPPORTING US

 $\mathcal{E}_{C,G,E,M}^{Centre of Genomics, Evolution & Medicine}$

The NSHG-PM 2023 workshop is organized together with Centre for Genomics, Evolution and Medicine (cGEM) of Institute of Genomics, University of Tartu (European Union's Horizon 2020 grant no 810645).

For further information please visit https://cgem.ut.ee/



The Joint Committee of the Nordic Medical Research Councils (NOS-M) is a collaborating body for the Nordic research councils that finance medical research and is a sponsor of the NSHG-PM 2023 workshop in Tallinn.

For further information please visit: https://nos-m.org/

NSHG-PM 2023

HTTPS://NSHG-PM2023.ORG

WORKSHOP

INDEX

Thanks for supporting us	2
Join NSHG-PM	4
Welcome	5
General Information	7
Programme	. 9
Floor Plan	.15
Poster Overview	.16
Industry Directory	19

INFORMATION

JOIN NSHG-PM

The Nordic Society of Human Genetics and Precision Medicine (NSHG-PM) represents a rapidly growing movement of scientists, clinicians, and other stakeholders who are demonstrating the value of combining information from genetics, -omics, imaging, and a variety of health and other registry data, with the goal of more precision in disease prevention and treatment. In order to achieve this, we need to build bridges from research to public health and clinical implementation of precision medicine.

Regular membership is open to researchers, clinicians, and others working on precision medicine research, translation, infrastructure, and practice in the Nordic countries. Trainee membership is available for advanced-degree precision medicine professionals during fellowships, as well as students working toward a post-baccalaureate or bachelor's degree.

1-Year MembershipEUR 501-Year Membership - Trainee/StudentEUR 30

For more information please visit www.nshg-pm.org/membership

NSHG-PM Council

Ole Andreassen President Norway

Ingrid Kockum Vice-President Sweden

Søren Brunak Treasurer Denmark

Bo Jacobsson Sweden Andres Metspalu

Lili Milani Estonia Pål Njølstad

Norway

Aarno Palotie Finland

Samuli Ripatti Finland

Contact

Hakon Heimer Executive Officer hakon.heimer@nshg-pm.org Jan Kristensen Secretariat Manager jnk@cap-partner.eu Hreinn Stefánsson

Kári Stefánsson Iceland

Thomas Werge Denmark WELCOME

We are delighted to see you join us in the historic city of Tallinn this summer for an enriching academic event. This gathering is brought to life through the concerted efforts of the Nordic Society of Human Genetics and Precision Medicine (NSHG-PM) and the University of Tartu.

Our program is meticulously designed to foster an environment ripe with stimulating discussions aimed at propelling Nordic precision medicine forward. Our organizing committee has arranged an agenda featuring over 20 insightful talks that delve into a range of pertinent subjects. These include the implementation of polygenic risk scores (PRS) and whole-genome sequencing in the clinic, potential applications of pharmacogenomics, machine learning, and custom disease risk scoring tools, among others.

This event holds a special focus on the young researchers and clinicians who are shaping the future of our scientific landscape. To this end, we have introduced workshops to this year's NSHG-PM annual meeting, addressing pivotal topics for the future of personalized medicine, including machine learning, Mendelian randomization, and PRS. Furthermore, we have organized a poster session, providing a platform for at least 25 young researchers to present their latest findings. The most promising of these will have the opportunity to deliver short talks to the auditorium.

We envision this workshop as a steppingstone towards establishing a framework that will fuel future public health, clinical care, and innovation in the Nordic region.

Join us in Tallinn for this enriching experience and seize the unique opportunity to network with fellow human geneticists, big data scientists, biobankers, epidemiologists, hospital clinicians, biomedical industry researchers, and executives.

Looking forward to seeing you in Tallinn!

On behalf of the Local Organizing Committee:



Lili Milani Head of Estonian Genome Centre Institute of Genomics, University of Tartu

Andres Metspalu Head of Estonian Biobank, Institute of Genomics, University of Tartu



Jon Anders Eriksson Head of Centre for Genomics, Evolution and Medicine, Institute of Genomics, University of Tartu



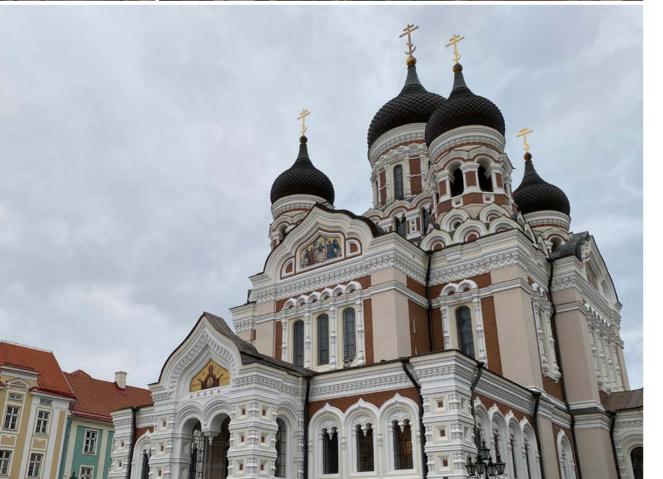
Erik Abner Estonian Genome Centre, Institute of Genomics, University of Tartu



Genetic and Personalized Medicine Clinic & Eye Clinic, Tartu University Hospital, Department of Clinical Genetics, Institute of Clinical Medicine, University of Tartu







GENERAL INFORMATION

Contact

NSHG-PM Secretariat CAP Partner Nordre Fasanvej 113, 1st floor 2000 Frederiksberg C, Denmark T: +45 70 20 03 05 info@cap-partner.eu www.nshg-pm.org

Conference Venue

Swissotel Tallinn Tornimae Street 3 10145 Tallinn, Estonia

Follow us on social media

in y

Search for NSHGPM Use #NSHGPM2023 when posting about the workshop!

Abstract Book

An electronic abstract book is available online at: nshq-pm2023.org/posters-and-abstracts

Certificates of Attendance

Certificates of attendance will be available as self-print after the conference. A link will be provided by e-mail to all participants.

Lunch, coffee and buffets

Lunch and coffee are available in the exhibition area. See programme for exact time of breaks. An evening networking buffet as well as a light poster session buffet will also be set up in the area. All food and beverages are included in the registration fee.

Speaker Information

Please bring your presentation to the conference room before your session begins. We recommend you upload your presentation in a break before your session starts. A technician will be present to assist in the upload if necessary. Please bring your presentation on a USB. Use of personal laptops is not allowed. Unless otherwise agreed all presentations will be deleted after the conference to make sure no copyright issues may arise.

Posters

Posters are to be mounted on Monday 19 June from 15.00-16.00 and must be removed by the end of the poster session on Tuesday 20 June. The measurement of your poster should be A0 Portrait.

The NSHG-PM Conference Secretariat provides equipment for setting up the posters on the wall or boards. The conference secretariat takes no responsibility for left or damaged posters.

Wi-fi

The Wi-fi code at Swissotel is: events.

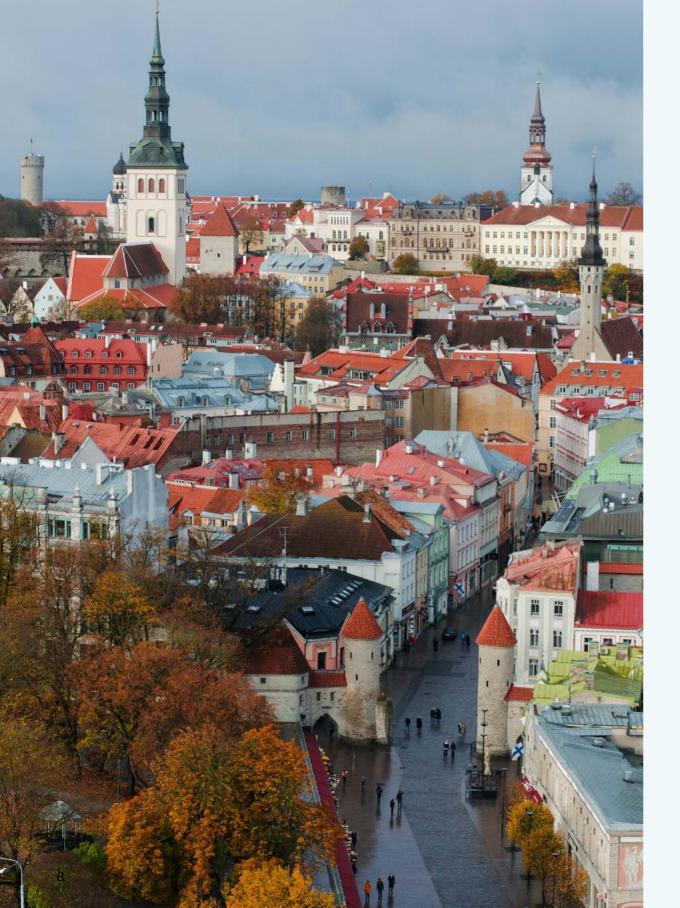
Mobile phones

All mobile phones must be on silent mode during the sessions. We encorage you to share pictures and experinces from the conference with colleagues - both in person and on social media, but please show consideration for the people in your photographs when you share them.

Free public transport

The city of Tallinn has offered to sponsor free public transport during the workshop days from 18-22 June 2023 for our participants. The tickets are valid on Tallinn bus lines nr 1-73, trolleybuses, and trams.

Please find your ticket and more information here: nshg-pm2023.org/venue INDUSTRY



Monday, 19 June 2023

15.00-16.00	Welcome & registration
16.00-16.15	Welcome from organizers: Lili Milani, University of Tartu, Estonia
16.15-17.00	KEYNOTE: Andrew McIntosh, University of Edinburgh, Scotland "Key insights from genomic studies of depression"
17.00-17.30	Coffee & snacks
17.30-19.00	CARDIOMETABOLIC SESSION Moderator: Maris Alver, Institute of Genomics, University of Tartu Talk 1: Jacob Bergstedt, Karolinska instituttet, Sweden: "Shared genetic liability, risk factors, and causal effects explaining the association between major depressive disorder and cardiovascular disease"
	Talk 2: Samuel Jones, University of Helsinki, Finland "Circadian GWAS of glucose and vitamin D identifies time-dependent genetic effects""
	Talk 3: Tove Fall, Department of Medical Sciences, Uppsala University, Sweden: "SCAPIS, a new resource of detailed data for precision medicine research"
	Talk 4: Dennis van der Meer, NORMENT, University of Oslo, Norway: "Genome-wide association study of MRI-derived measures of body composition reveals genetic overlap with cardiometabolic traits and disorders"
19.00-21.00	Networking and evening buffet

INFORMATION

PROGRAMME

FLOORPLAN

POSTER OVERVIEW

INDUSTRY

NSHG-PM 2024

08.00-09.00	Welcome registration & morning coffee		15.30-16.00	Coffee & snacks
09:00-10.30	METHODS FOR INTERVENTION		16.00-17.00	Technology session
	Moderator: Marc Vaudel, University of Bergen, Norway			Moderator: Tõnu Esko, University of Tartu, Estonia
	Talk 1: Elise Koch, CoE NORMENT, University of Oslo, Norway: "Genomic Prediction of Antipsychotic Prescription Patterns"			Cindy Lawley, Director, Population Health, Olink: "Accelerating translational research at population scale, enabling biobanks and clinical cohorts as valuable tools for precision medicine"
	Talk 2: John Shorter, Roskilde University, Denmark: "The performance and implementation of PRS from the trans-Nordic collaboration on major depressive disorder"			Sini Kerminen, Senior Data Scientist, Nightingale Health: "Population scale blood biomarkers for preventative health"
	Talk 3: Hreinn Stefansson, deCODE genetics, Iceland: "Variants conferring high-risk of schizophrenia affect cognition in controls"			Shantanu Kaushikkar, Head of Business Development, Thermo Fisher: "Predictive genomics: harnessing the genome to prevent disease"
	Talk 4: Pekka Vartiainen, University of Helsinki, Finland: "Risk factors and clinical prediction model for severe RSV infection in infants"			Danielle Folkard, Sales Director, EMEA North, Oxford Nanopore: "What you're missing matters"
10.30-11.00	Coffee & snacks		17.00-19.00	Poster session short talks:
11.00-12.30	ENVIRONMENT EFFECTS ON HEALTH Moderator: Ingrid Kockum, Karolinska Institute, Sweden Talk 1: Ingibjörg Magnusdottir, Centre of Public Health Sciences,			Talk 1: Pernilla Stidh, Karolinska Institutet, Sweden: "A novel locus associated with multiple sclerosis severity implicates CNS resilience in disease outcome"
	University of Iceland: "Mental, Physical and Cognitive manifestations of the post COVID-19 condition: Results from the international COVIDMENT consortium"		Talk 2: Mischa Lundberg, Institute of Biological Psychiatry, Denmark: "Incorporating family history in GWAS of Major Depressive Disorder improves detection of polygenic signal"	
	Talk 2: Triin Laisk, University of Tartu, Estonia: "Genetics of cervical cancer and high-risk HPV infection"			Talk 3: Kira Detrois, Institute for Molecular Medicine Finland (FIMM), Finland "Polygenic risk scores enhance onset prediction for 7/18 diseases when added to models with environmental and clinical risk factors"
	Talk 3: Lars Alfredsson, Karolinske Institutet, Sweden: "Airborne environmental exposure and risk of MS and RA"			Talk 4: Karolina Anja, University of Tartu, Estonia: "The genetic basis of eating behaviors"
	Talk 4: Jon Anders Eriksson, University of Tartu, Estonia: "Gene-environment interactions in metabolic traits: challenges and opportunities for personalised medicine"			Talk 5: Ingunn Olea Lund, Norwegian Institute of Public Health, Norway: "Prenatal maternal stress: triangulating evidence for intrauterine exposure effects on birth and early childhood outcomes across multiple approaches"
12.30-13.30	Lunch			
13.30-15.30	USE OF POLYGENIC RISK SCORES IN SCIENCE AND CLINIC Introduction by: Kari Stefannson, deCODE genetics, Iceland (TBC)			Talk 6: Eero Aaltonen, Institute for Molecular Medicine Finland (FIMM), Finland: "Integrating a chat-based doctor's appointment to KardioKompassi encourages preventive action in individuals with elevated risk of coronary artery disease."
	Speakers: Joeri Meijsen, Institute of Biological Psychiatry (IBP), Sct. Hans, Denmark: "Fundamentals of Polygenic Risk Scores"			+ Presentations in poster exhibition including light snacks
	Kristi Läll, University of Tartu, Estonia: "Pitfalls and opportunities of polygenic risk scores"			
	Elisabeth Widen, University of Helsinki, Finland: "Polygenic Risk Scores - Potential to Impact Clinical Care"			
	Neeme Tõnisson, University of Tartu, Estonia: "Polygenic risk scores in Estonian studies with research participants and beyond"			
	Round-table discussion Moderator: Kristi Läll, University of Tartu, Estonia			

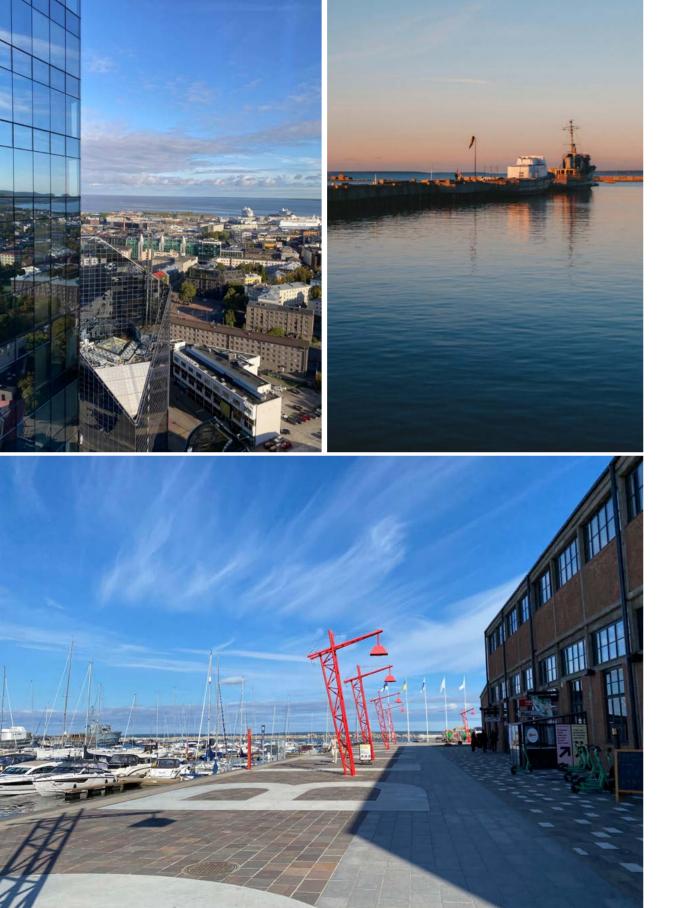
11

08.30-09.00	Welcome & morning coffee		
09.00-09.30	 Workshop intro Workshops: A: Polygenic risk scores B: Mendelian randomization C: AI & Machine learning Participants must choose one of the three workshops 		
	Polygenic risk scores Co-Leaders: John Shorter, Roskilde University, Denmark Joeri Meijsen, Institute of Biological Psychiatry, Denmark	Mendelian randomisation Co-Leaders: Joëlle Pasman, Karolinska Institutet, Sweden Guðmundur Einarsson, deCODE genetics, Iceland	Al & Machine Learning Co-Leaders: Anders Dale, University of California, San Diego, USA Hafsteinn Einarsson, deCODE genetics, Iceland
11.00-11.30	Coffee & snacks		

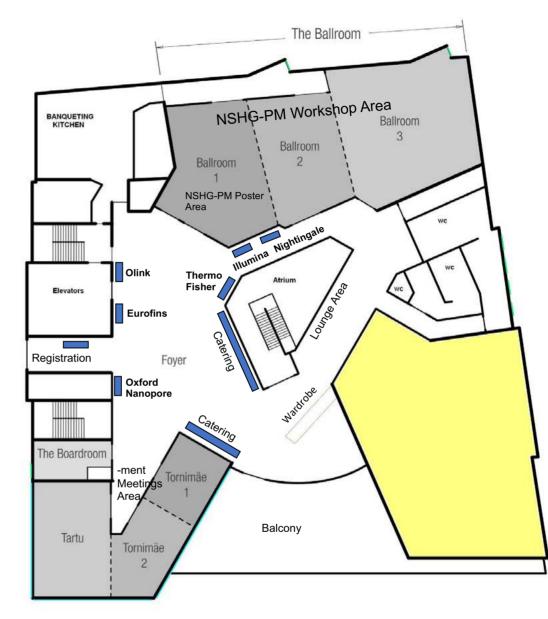
11.30-12.30	Workshop A	Workshop B	Workshop C
	Polygenic risk scores	Mendelian randomisation	AI & Machine Learning
	The purpose of this workshop is to understand the strengths and limitations of polygenic scores through hands-on exercises. Participants will work with two different tools to better understand polygenic scores. This includes the PGS Catalog, which is an open database of published polygenic scores, and LDpred2, which is a method for deriving polygenic scores from GWAS summary statistics. No previous experience with coding is needed for the exercise with the PGS Catalog. Experience with the programming language R would be helpful (but not required) for the LDpred2 exercise. By the end of the workshop, participants will: understand how PGS are created, how to evaluate their predictive performance, and how to search for performance differences across different ancestry groups.	In this workshop, you will learn the theoretical basis and practical skills needed to understand and conduct Mendelian Randomization (MR). You will learn to use the webtool MR Base as well as conduct your own analyses in R, using real GWAS summary statistics to test the causal relationship between your favorite disease traits. After this workshop, you will be able to evaluate the bold causal claims often made by studies using MR, and you will have the skills and scripts to conduct basic MR analyses yourself.	The advent of new machine learning approaches has led to numerous real-world applications, particularly in the healthcare sector. Techniques such as image analysis and pattern recognition have become increasingly relevant in practice, enhancing diagnostic capabilities and patient care. When combined with genetics, these advancements have the potential to usher in a new era of personalized medicine, transforming the way we understand and treat a myriad of health conditions. To better understand the potential of this new approach, we invite you to participate in the machine learning workshop.
12.30-13.30	Lunch		
13.30-14.30	Workshop A	Workshop B	Workshop C
	Polygenic risk scores	Mendelian randomisation	AI & Machine Learning
14.30-15.00	Coffee & snacks		
15.00-16.00	Summary session & closin	g event	

Wednesday, 21 June 2023

13



FLOOR PLAN



POSTER OVERVIEW

 Hormonal imbalance and risk of multiple sclerosis in women: findings from a nationwide cohort study and genome-wide cross-trait analysis Shared genetic architecture between schizophrenia and anorexia nervosa: a cross-trait genome-wide analysis Using common genetic variance to study Copy Number Variations (CNVs) role in complex human traits Rare copy number variation in Addison's disease Genome-wide association study reveals a locus in ADARB2 gene for complete freedom from headache Yean Jiang Yean Jiang Yean Jiang Yean Jiang Yean Jiang Sean Jone Study and study and study reveals a locus in hadache 	Snihirova aza
 and anorexia nervosa: a cross-trait genome-wide analysis 3 Using common genetic variance to study Copy Number Variations (CNVs) role in complex human traits 4 Rare copy number variation in Addison's disease Haydee Arta 5 Genome-wide association study reveals a locus in 	Snihirova aza
Number Variations (CNVs) role in complex human traits4475669910<	aza
5 Genome-wide association study reveals a locus in Isa Olofssor	
,	١
6 Polygenic risk scores enhance onset prediction Kira Detrois for 7/18 diseases when added to models with environmental and clinical risk factors	
 7 A map of genetic and phenotypic associations Jéssica Figuracross male reproductive phenotypes 	Jerêdo
8 A DNA Methylation Study of Common and Specific Jonelle Villa Treatment Effects of Three Antipsychotics in the Treatment of Psychosis	ır
 Integrating a chat-based doctor's appointment to KardioKompassi encourages preventive action in individuals with elevated risk of coronary artery disease 	en
10COVID-19 and risk of mental health problems: A mendelian randomization study using data from the Norwegian Mother, Father and Child Cohort StudyBin Wang	
11 Risperidone treatment reverts differences in estimated blood cell proportions and DNA methylation in antipsychotic-free first episode of psychosisLeticia Spin	dola
12 Quantifying The Assortative Mating of Psychiatric Saeid Rasek Disorders and its Consequences in Danish Population	khi Dehkordi
13 Genetic associations with age of rheumatoid arthritis Karina Patas	sova
14 Quantifying the relative importance of genetics and Joeri Meijse the environment on the comorbidity between mental and cardiovascular disorders using register data	en

No.	Title	Presenter
15	Repeats R/T ratio is associated with Human Y chromosome Haplogroups	Tarmo Puurand
16	Investigating the causal effects of childhood and adulthood adiposity on later life mental health outcome: a Mendelian randomisation study	Sweta Pathak
17	Polygenic scores for psychiatric disorders associate with year of first bipolar disorder diagnosis: a register-based study between 1972 and 2016	Lina Jonsson
18	Mutational analysis profile in patients with myeloproliferative neoplasms in relation to hemostatic complications – a retrospective study	Ines Vaide
19	Preliminary Genome-Wide Analysis of Self-Reported Psychotic Experiences in the Norwegian Mother, Father, and Child study	Viktoria Birkenæs
20	Personality traits predict vaccination against COVID-19 better than a polygenic score and demographics	Uku Vainik
21	Eating problems among adolescents before and during the Covid-19 pandemic	Johanne Pettersen
22	Prenatal maternal stress: triangulating evidence for intrauterine exposure effects on birth and early childhood outcomes across multiple approaches	Ingunn Olea Lund
23	Incorporating family history in GWAS of Major Depressive Disorder improves detection of polygenic signal.	Mischa Lundberg
24	A novel locus associated with multiple sclerosis severity implicates CNS resilience in disease outcome	Pernilla Stridh
25	Explaining familial risk of diseases with interpretable unsupervised latent representations from comprehensive nationwide Finnish data	Tuomo Hartonen
26	The genetic basis of eating behaviors	Karolina Anja
27	Phenome-wide association study of ADHD genetic liability and ICD-10 medical conditions in the Estonian Biobank	Elis Haan

V Olink

Empower your genomics with proteomics

- Discover novel drug targets
- Reveal actionable insights into disease pathways
- Bridge the genotype-phenotype gap

To learn more, please scan



illumina

Unlock the power of genomics in precision health

Improving health management through genomics-powered precision medicine. Catch up with our recent Preventative Genomics event talks now:





INDUSTRY DIRECTORY

Gold Sponsors

	Eurofin Genomics www.eurofinsgenomics.com
eurofins Genomics	FAST. We produce and analyse DNA & RNA COMPLETE. We support from research to application RELIABLE. We work with pharma, food, diagnostics, agriculture & biotechnology Learn more: eurofinsgenomics.com
illumına [*]	Illumina www.illumina.com Illumina is a leading developer, manufacturer, and marketer of life science tools and systems to make genomics useful for all. We reimagine what's possible for human health, including how diseases are discovered, detected, diagnosed, and treated. Our sequencing innovations enable researchers and clinicians to accelerate the future of personalized medicine.
Olink [®]	Olink www.olink.com Olink offers an unmatched high-multiplex technique to identify actionable biomarkers, with a strong focus on the human plasma proteome. Using minimal sample volume we provide quantifiable results with high-throughput, exceptional sensitivity and specificity, with coverage across a broad dynamic range.
	Oxford Nanopore www.nanoporetech.com Oxford Nanopore Technologies has developed the world's first and only nanopore DNA/RNA sequencing devices, scalable to your requirements. Oxford Nanopore brings real-time sequencing with unrestricted read lengths, to anyone, anywhere, whether in scientific research, education, or real-world applications: from clinical research and population genomics to outbreak surveillance and environmental monitoring.
Thermo Fisher S C I E N T I F I C The world leader in serving science	Thermo Fisher www.thermofisher.com Thermo Fisher Scientific is the world leader in serving science. Customers worldwide trust our tools, services, and solutions to help them accelerate innovation and enhance productivity. Together, we are making advancements that make a real difference. Our Mission is to enable our customers to make the world healthier, cleaner, and safer.

Silver Sponsor



Nightingale www.nightingalehealth.com

Nightingale Health offers next-generation clinical chemistry for research Replicate and publish your research findings, adding to 450+ prior publications from UK Biobank and other cohorts.

Identify causative mutations in unsolved genetic diseases

- Explore large and small variants, methylation, and phasing
- Span and quantify repeat expansions
- Rapid access to results



Anything • Anyone • Anywhere



visit • nanoporetech.com follow • @nanopore

Oxford Nanopore Technologies and the Wheel icon are registered trademarks of Oxford Nanopore Technologies pic in various countries. © 2023 Oxford Nanopore Technologies pic. All rights reserved. Oxford Nanopore Technologies products are not intended for use for health assessment or to diagnose, treat, mitigate, cure, or prevent any disease or condition.

ThermoFisher

Total solution.

Moving toward better outcomes for you and better treatment for all.

Work to advance your predictive genomics goals with solutions for every step, from sample storage and analysis and reporting.





applied biosystems

Learn more at thermofisher.com/predictivegenomics

For Research Use Only. Not for use in diagnostic procedures. © 2023 Thermo Fisher Scientific Inc. All rights reserved. All trademarks are the property of Thermo Fisher Scientific and its subsidiaries unless otherwise specified COL023853 0523

NOTES

NSH-PM 2023 Workshop

EUROFINS GENOMICS THE DNA UNIVERSE

FAST. We produce and analyse DNA & RNA. Eurofins Genomics is part of the Eurofins Group, with facilities in Europe, the USA and Asia. We are an internationally leading provider of DNA sequencing services, genotyping services, DNA synthesis products and bioinformatics services.

COMPLETE. We support you from research to application. With our assistance, you will master any challenge along your value chain. We combine outstanding expertise with state-of-the-art instruments, methods and technologies. We support standard and challenging research projects, as well as complex strategies for large interdisciplinary companies.

RELIABLE. We work with academics, pharma, food, diagnostics, agriculture and biotechnology. Our highly automated and lean-optimised lab processes allow us to deliver highly-reliable results. This is documented by the ISO 17025 and ISO 13485 accreditation, as well as the GLP, GCP and cGMP certifications.

Reach for the stars. We are glad to assist you! Learn more at www.eurofinsgenomics.com

EUROFINS GENOMICS Viehmarktgasse 1B, Büro 2 / 1030 Vienna, Austria support-eu@genomics.eurofinseu.com / eurofinsgenomics.com / +43 1 7966572



🔅 eurofins

BIOBANKS, RESEARCH, AND PRECISION MEDICINE A Joint Nordic Conference on Future Health

September 10-12, 2024 * Trondheim, Norway





Tallinn

Thanks to the city of Tallinn for sponsoring free public transport during the workshop days from 18-22 June 2023 for our participants.

Please find the ticket here: https://nshg-pm2023.org/venue

Mark your calendars for a gathering of the Nordic precision health community!

Next year, the biobank and research communities will join forces for a special Nordic-wide community gathering to explore the present and future of precision health. Make plans now to join us September 10-13, 2024, in lovely Trondheim, Norway, for:

- Overviews of precision prevention, diagnosis, and treatment in the Nordics
- Research symposia covering the breadth of cutting-edge research
- Critical discussion of the issues important to Nordic biobanks
 - O Funding priorities
 - O Legal and ethical considerations
 - o Citizen involvement
- Policy discussions about the best way forward

We anticipate more than 700 participants and invite you to contact us early with ideas. We welcome partners and sponsors from all sectors that have a stake in the successful development of a cooperative Nordic precison health future: policy organizations, health systems, research entities, industry, as well as journals and other media.

Contacts: Nordic Biobank Network, Kristian Hveem (kristian.hveem@ntnu.no) or NSHG-PM Jan Kristensen (jnk@cap-partner.eu) to learn more.

THANK YOU TO OUR SPONSORS

GOLD SPONSOR



WWW.NSHG-PM2023.ORG