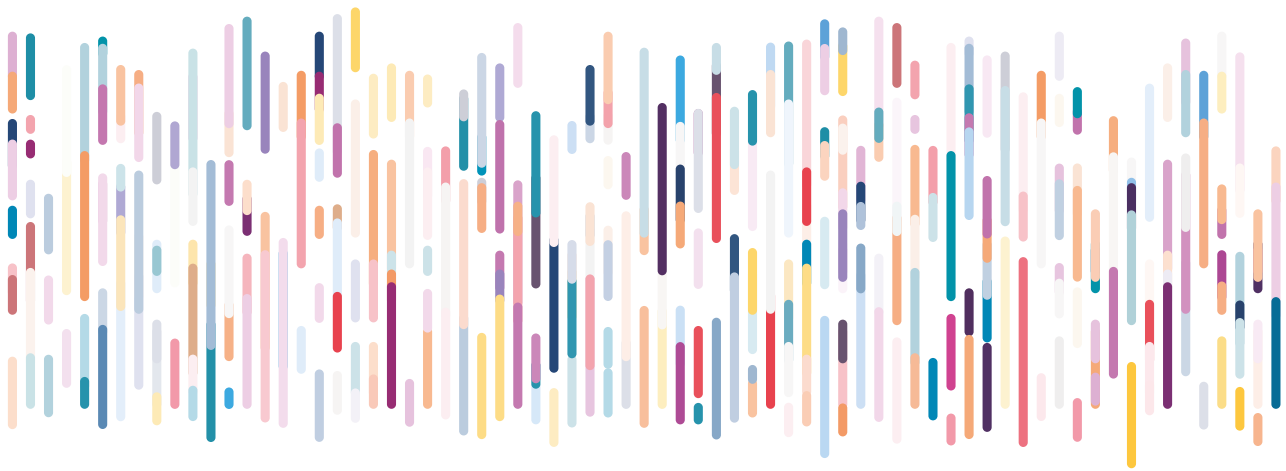
2025 Gene Forum

The 24th Annual International

9–10 September 2025 Tartu, Estonia



UNIVERSITY OF TARTU
Institute of Genomics

Eesti Geenikeskus
Estonian Genome Foundation

Funded by
the European Union

WELCOME

It is with great pleasure that we welcome you to the 24th International Gene Forum, held in the historic city of Tartu at the Estonian National Museum.

Since its inception, Gene Forum has become the leading platform for scientific exchange in genetics and genomics across the Baltic region and beyond. Over the years, the forum has hosted more than 400 renowned speakers from 34 countries, fostering dialogue that has advanced both fundamental research and clinical applications.

This year's programme reflects the rapidly evolving landscape of genomics, with sessions dedicated to complex genomic approaches, reproductive genetics, disease mechanisms, ageing, and the convergence of medical genetics with big data. We are also excited to explore topics at the intersection of basic science and human history, including human origins and the microbiome.

As always, Gene Forum provides an unique opportunity for researchers, clinicians, and industry leaders to connect, collaborate, and push the boundaries of our shared knowledge.

We hope that the conference will inspire fresh ideas, foster lasting collaborations, and continue to serve as a catalyst for innovation in genetics and genomics.

On behalf of the Gene Forum 2025 Programme Committee
Mait Metspalu

Mait Metspalu
Institute of Genomics,
University of Tartu, Director of Institute of Genomics,
Professor of Evolutionary Genomics

Andres Metspalu
Institute of Genomics,
University of Tartu,
Professor of Genomics and Biobanking, Academician

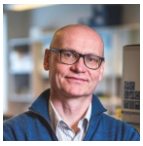
Lili Milani
Institute of Genomics,
University of Tartu,
Head of Estonian Biobank,
Professor of Epi- and Pharmacogenomics

Elin Org
Institute of Genomics,
University of Tartu, Deputy Director of Estonian Genome Centre,
Professor of Microbiomics

Triin Laisk
Institute of Genomics, University of Tartu,
Associate Professor of Genomics and Reproductive Genetics

Michael Dannemann
Institute of Genomics, University of Tartu,
Associate Professor of Evolutionary and Population Genomics, Head of Centre for Genomics, Evolution and Medicine

Sander Pajusalu
Faculty of Medicine, University of Tartu,
Vice Dean, Associate Professor of Clinical Genetics, Tartu University Hospital, Head of the Genetics and Personalized Medicine Clinic



Pärt Peterson
University of Tartu, Faculty of Medicine,
Institute of Biomedicine and Translational Medicine,
Professor of Molecular Immunology

Kaur Alasoo
University of Tartu, Faculty of Science and Technology, Institute of Computer Science, Associate professor of Bioinformatics

GENERAL INFORMATION

Conference venue

Estonian National Museum

Muuseumi tee 2
60532, Tartu, Estonia

Organisers

University of Tartu, Institute of Genomics

Riia 23b,
51010, Tartu, Estonia
e-mail: geneforum@ut.ee

Estonian Genome Foundation

Tiigi 61b,
50410, Tartu, Estonia

Meet our organizing committee

Kaisa Lankots
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Abstract book

An electronic abstract book of poster presentations is available at https://geneforum.ee/poster-session/

You can find more information on - our invited speakers and their presentations here

Certificates of Attendance

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

Certificates of Attendance

Poster session takes place on the 9th of September starting from 16:20 with 4 poster-talks selected by the programme committee and followed by a joint poster viewing in the poster area from 17:00. Posters have been divided into 6 categories and are set up in the Ilmar Manninen auditorium and its entrance.

An electronic abstract book of poster presentations is available at https://geneforum.ee/poster-session/

Please do not take pictures of posters that have the restricted marking.

Networking event

The networking event of Gene Forum 2025 takes place on the 9th of September starting from 19:30 in the atrium before the museum exhibition entrance. In order to participate in the networking event, please be sure you have purchased a ticket including the additional programme.

Guided tour of museum

You are welcome to join a guided tour of the Estonian National Museum on 9th September at 18:00. For more details on available options, please contact the organisers. Kindly note that participants are responsible for purchasing their own museum/exhibition tickets.

Mobile phones

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

Gene Forum 2024 is held as Ethical MedTech Conference Vetting System approved event

For more information visit our website www.geneforum.ee

PROGRAMME

Tuesday, September 9th, 2025

8:30–9:00 REGISTRATION

9:00–9:15 OPENING OF THE CONFERENCE

Kristina Kallas
Minister of Education and Research of Estonia

9:15–9:50 KEYNOTE

Eimear Kenny

Endowed Chair and Professor for Genomic Health at Icahn School of Medicine at Mount Sinai, USA
"Population Genetics in an Era of Genomic Health ".

9:50–10:45 COMPLEX GENOMIC APPROACHES

Moderator Lili Milani

Olli Pietiläinen

Group Leader and senior scientist, Helsinki Institute of Life Science, Neuroscience Center, University of Helsinki, Finland
"Using multimodal, longitudinal data to explore clinical impact of genetic variants in psychotic disorders"

Simone Rubinacci

FIMM-EMBL Group Leader at University of Helsinki and Broad Institute of MIT and Harvard, USA
"Haplotype-informed methods to uncover novel disease mechanisms"

10:45–11:20 COFFEE BREAK

11:20–12:40 REPRODUCTIVE GENETICS

Moderator Triin Laisk

Katherine Ruth

Lecturer in Clinical and Biomedical Sciences, University of Exeter, United Kingdom
"Insights from genome-wide analyses of female reproductive ageing"

Pol Sole-Navais

Group leader at the Wallenberg Laboratory, University of Gothenburg, Sweden
"Genetic susceptibility to shorter gestational duration and preterm birth"

Taru Tukiainen

Senior Researcher, University of Helsinki, Finland
"Genetic insights into the lifelong health of women and children"

PROGRAMME

Tuesday, September 9th, 2025

12:40–13:55 LUNCH BREAK

13:55–14:50 DISEASE MECHANISMS

Moderator Kaur Alasoo

Hilary Martin

Group Leader in Human Genetics, Wellcome Sanger Institute, United Kingdom

"Insights into the genetics of neurodevelopmental conditions and traits from large British cohorts"

Stephen Burgess

MRC Biostatistics Unit, Univeristy of Cambridge, United Kingdom

"Mendelian randomization: How can genetics guide the design of clinical trials?"

14:50–15:25 COFFEE BREAK

15:25–16:20 AGEING

Moderator Pärt Peterson

Juulia Jylhävä

Senior Research Fellow and Group leader at Tampere University, Finland and Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden

"Decoding biological aging and frailty through genomic methods"

Joris Deelen

Associate professor at Leiden University Medical Center, The Netherlands

"Metabolomics-based biomarkers of ageing; how to implement epidemiological findings into clinically relevant settings"

16:20–17:00 POSTER SESSION presentations

Moderator Triin Laisk

Valentina Rukins

Junior Research Fellow in Genomics, Institute of Genomics, University of Tartu, Estonia

"Multi-ancestry, trans-generational GWAS meta-analysis of gestational diabetes and glycaemic traits during pregnancy reveals limited evidence of pregnancy- specific genetic effects"

Maarja Jõeloo

Research Fellow in Structural Genomics, Institute of Genomics, University of Tartu, Estonia

"Characterisation of rare copy number variation effects on circulating metabolic biomarkers"

PROGRAMME

Tuesday, September 9th, 2025

16:20–17:00 POSTER SESSION presentations

Kateryna Pantiukh

Junior Research Fellow, Institute of Genomics, University of Tartu, Estonia

"Metagenome-assembled genomes from a population-based cohort uncover novel gut species and strain diversity, revealing prevalent disease associations"

Getachew Arage

Postdoctoral Researcher, Uppsala University, Sweden

"Association between meat intake and subclinical atherosclerosis in the population-based Swedish CardioPulmonary BioImage Study"

17:00– … POSTER SESSION

19:30– … DINNER AT ESTONIAN NATIONAL MUSEUM

PROGRAMME

Wednesday, September 10th, 2025

9:00–10:20 MEDICAL GENETICS MEETS BIG DATA

Moderator Sander Pajusalu

Nicola Whiffin
Associate Professor, Big Data Institute, University of Oxford, United Kingdom
"Small nuclear RNAs in rare disease"

Peter Krawitz
Professor at Institute for Genomic Statistics and Bioinformatics of the University Hospital Bonn and the Rheinische Friedrich-Wilhelms-Universität Bonn, Germany
"GestaltMatcher: medical image analysis with AI in rare diseases"

Aleš Maver
MD, PhD, Clinical institute of genomic medicine, UMCL Ljubljana, Slovenia
"The power of small – how Slovenia used genomics to transform diagnosis of genetic diseases"

10:20–10:55 COFFEE BREAK

10:55–12:15 TECHNOLOGICAL ADVANCES IN THE FOREFRONT OF SCIENCE

Moderator Lili Milani

Neil Ward
VP and General Manager of PacBio for Europe, the Middle East, and Africa
"Discovering new biology with HiFi"

Agnieszka Ciesielska
Senior Technical Sales Specialist, Element Biosciences
"High-Dimensional Biology: Advancing Single-Cell and Spatial Multiomics.with AVITI24TM"

David Parry
Staff Bioinformatics Scientist, Illumina
"How AI is powering the next wave of genomic discovery: Illumina AI Lab Innovation Roadmap"

PROGRAMME

Wednesday, September 10th, 2025

12:15–13:30 LUNCH BREAK

13:30–14:50 HUMAN ORIGINS

Moderator Michael Dannemann

Pontus Skoglund
The Francis Crick Institute, United Kingdom
"Ancient genome evolution in humans and bacteria"

Mari Sepp
Postdoctoral researcher, Heidelberg University, Germany
"Tracing cerebellum development and evolution with single-cell genomics"

Martin Kuhlwilm
Assistant Professor, Department of Evolutionary Anthropology, University of Vienna, Austria
"Understanding human uniqueness using hominin and primate genomic diversity"

14:50–15:25 COFFEE BREAK

15:25–16:45 MICROBIOME

Moderator Elin Org

Tove Fall
Professor of Molecular Epidemiology, Uppsala University, Sweden
"Genome-wide association study of the microbiome composition in four Nordic cohorts"

Rob Finn
Section Head & Senior Scientist, EMBL-EBI, UK
"Exploring microbial diversity across scales and biomes"

H Bjørn Nielsen
Chief Scientific Officer at Cmbio, Copenhagen, Denmark
"Status and Perspective on the Microbiome Field"

16:45–17:20 CLOSING REMARKS

Announcing the winner of Artur Lind Scholarship

POSTER OVERVIEW

	EVOLUTION AND POPULATION GENETICS	Presenter
1	The Underlying Evolutionary Dynamics of the Human Immune Response	Rutvi Rajpara
2	The conundrum of nc886 – methylation pattern with clear association to ancestral origins, but no genetic determinants?	Saara Marttila
3	Sperm competition intensifies purifying selection on testis-specific genes in primates	Vasili Pankratov
4	Application of spherical variational autoencoders to human population genetic structure	Elia Tiso
5	Signatures of selection and adaptation in an Arctic Circle population	Rodrigo Flores
	GENETICS OF HUMAN TRAITS AND DISEASES	
6	GWAS reveals importance of vaginal epithelium associated genes in case of recurrent vaginitis	Evelin Mutli, Reet Mändar
7	Music style preferences and well-being: A genetic perspective	Anastasiia Bratchenko
8	Genetic architecture of the Big Five personality traits and their associations with health behaviours	Kerli Ilves
9	Integrating brain structure and function for the neurobiology and genetics of language	J.S. Amelink
10	Genetic non-additivity reveals variants in complex traits plasticity	Ralph Porneso
11	Association between plausible genetic factors and weight loss from GLP1-RA and bariatric surgery	Uku vainik
12	GWAS Meta-Analysis of ADHD Symptoms in Adults Reveals Differential Genetic Architecture of Inattention and Hyperactivity-Impulsivity Domains	Triinu Varvas
13	Coding variant analysis based on 5,522 ICD-10-based disease phenotypes in the Estonian biobank	Kanwal Batool
14	Recessive variants in the DARS2 gene as a novel cause of axonal Charcot-Marie-Tooth disease	Siiri Sarv
15	Interpreting artificial neural networks to detect genome-wide association signals for complex traits	Burak Yelmen
16	Leveraging primary health care data from Estonian Biobank to discover novel genetic associations	Anastasiia Alekseienko
17	**Characterisation of rare copy number variation effects on circulating metabolic biomarkers**	**Maarja Jõeloo** **Poster-talk**
18	Text-based approach for detecting cases of ADEs from EHRs of participants of the Estonian Biobank	Dage Särg
19	Exploring the Estonian Gut Microbiota: Composition, Structure, and Research Potentia	Hindrek Teder
20	**Metagenome-assembled genomes from a population-based cohort uncover novel gut species and strain diversity, revealing prevalent disease associations**	**Kateryna Pantiukh** **Poster-talk**
21	A Bioconductor workflow for microbiome-based survival analysis	Sneha Das
22	Probabilistic alternatives for microbiome research in R/Bioconductor	Rasmus Hindström
23	Predictive markers of future depression in the gut microbiome	Annabel Klemets
24	Modeling geographic variation of human microbiome	João Paulo Cassucci dos Santos
25	Global antimicrobial resistance patterns in human gut metagenomes are structured along socio-economic gradients	Mahkameh Salehi
26	Biome-specific genome catalogues reveal functional potential of shallow sequencing	Dattatray Mongad

	REPRODUCTIVE GENETICS	Presenter
27	Leveraging genome-wide association study summary statistics to identify possible drug targets for uterine fibroids	Lisette Haug
28	Maternal Mood Disorders Are Associated With Increased Proportion of Placental Immune Cell Types Driving Gene Expression Differences	Soile Hytti
29	**Multi-ancestry, trans-generational GWAS meta-analysis of gestational diabetes and glycaemic traits during pregnancy reveals limited evidence of pregnancy-specific genetic effects**	**Valentina Rukins** **Poster-talk**
30	Genetic risk factors of vasomotor symptoms	Mia Golob
	RISK & PREDICTION	
31	Multi-omic risk prediction scores for rheumatoid arthritis in the Estonian Biobank	Galadriel Velázquez
32	No evidence for a causal effect of thyroid function and disease on risk for attention-deficit/hyperactivity disorder: A two-sample Mendelian randomization study	Triinu Peters
33	PREV-GEN – PREVentative outreach with GENetic testing	Kalle Pärn
34	**Association between meat intake and subclinical atherosclerosis in the population-based Swedish CardioPulmonary BioImage Study**	**Getachew Arage** **Poster-talk**
35	Integrating Polygenic Risk Scores into OMOP Common Data Model using the Estonian Biobank: A Preliminary Study of AI-Driven Cardiovascular Disease Prediction in Atherosclerosis	Djeane Debora Onthoni
36	Identifying predictors of weight loss from a diverse set of biological, behavioural, and psychological factors	Birgit Malken
37	Predicting blood metabolite profiles using multi-target neural networks	Merve Nur Güler
38	CVDLINK: Federated AI for Advancing Predictive Models of Cardiovascular Disease	Urmo Võsa
	PERSPECTIVE ON ADVANCED GENOMIC TECHNOLOGIES	
39	EASIGEN-DS: Designing a distributed Research Infrastructure on Advanced Genomics Technologies	Mireia Vaca-Dempere
40	The Genome of Europe: A Reference Dataset of Genomes	Andres Metspalu, Merit Kreitsberg

NOTES

NOTES

Artur Lind Scholarship

Artur Lind

Artur Lind (1927-1989) was an Estonian molecular biologist, who is also considered the founder of this field in Estonia. He studied protein biosynthesis, molecular biology methods, the structure of ribonucleic acid, methods for determining the sequence of deoxyribonucleic acid, oncogenes, and the structure and function of the ribosome, including being the first to isolate low-molecular-weight ribonucleic acid (5S-RNA) from eukaryotic ribosomes. Many of today's Estonian top scientists in the fieldhave worked and studied under him, such as Richard Villems, Mart Saarma, Mart Ustav, Toivo Maimets and Andres Metspalu.

To honour Artur Lind's significant contribution to Estonian science, the Estonian Genome Foundation established the Artur Lind Scholarship Fund in 2001. After a brief hiatus, the Institute of Genomics of University of Tartu, in collaboration with the Estonian Genome Foundation, is pleased to once again invite all doctoral students studying in Estonia, whose work is related to biomedicine, biotechnology, genomics, molecular biology, and adjacent fields, to apply for the scholarship.

In 2025, to apply for the scholarship doctoral students were asked to present a 3–5-minute video on the topic **"Human and Artificial Intelligence in Science in 2035"**

The scholarship recipients will be announced at the closing ceremony of the Gene Forum. The videos by the winners will be made public on our website after the conference.

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At **Illumina**, our mission is to improve human health by unlocking the power of the genome. Our sequencing by synthesis chemistry is used to generate high-accuracy DNA and RNA sequence data in studies around the globe. The innovative products that we provide are helping drive advancements in a multiomic understanding of cellular functions, combining transcriptomics, epigenetics, and proteomics. Multiomics profiling studies enable a more comprehensive understanding of development, cellular response, and disease, fueling the discovery of novel drug targets and biomarkers. The progress our customers have made and what lies ahead inspire us to push the boundaries of what is possible so we can create the next generation of genomics solutions.

Interlux is a leading provider of advanced solutions for the medical, scientific, and biotechnology sectors in the Baltics, and the authorized distributor of **Element Biosciences**. At Gene Forum, we are proud to showcase Element—a cutting-edge multiomics innovator. AVITI System combines high data quality, flexibility, and cost efficiency, empowering researchers to push the boundaries of genomics.

Visit our websites at: https://www.interlux.ee/ and https://www.elementbiosciences.com/

PacBio is a premier life science technology company that is designing, developing and manufacturing advanced sequencing solutions to help scientists and clinical researchers resolve genetically complex problems. PacBio products and technology under development stem from two highly differentiated core technologies focused on accuracy, quality and completeness which include our existing HiFi long read sequencing and our SBB® short read sequencing technologies. PacBio products address solutions across a broad set of research applications including human germline sequencing, plant and animal sciences, infectious disease and microbiology, cancer genomics, and other emerging applications.

SPONSORS

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BioNordika provides the tools for your research to succeed. We distribute premium life science brands, and offer products relevant for cell & molecular biology, immunology, diagnostics, instruments, and more!

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Genotipas Laboratory is your trusted partner in advanced genetic testing and personalized diagnostics. Our mission is to enhance lives through innovative, science-driven solutions for health, wellness, and performance.

At Genotipas, we offer a comprehensive range of genetic and molecular testing services to empower individuals and healthcare professionals with accurate, actionable insights for Cancer, Nutrition & Dermatology, Anti-Ageing, Healthy Lifestyle, Reproductive & Prenatal Health, Neonatal & Rare Disease Testing.

We simplify complex science to deliver results that matter - results you can trust

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HaploX is a global leader in precision NGS-based genetic testing, offering WES, WGS, and targeted panels for hereditary cancers, cardiovascular diseases, and rare disorders. Our solutions empower clinicians and researchers with actionable insights for diagnosis and family risk management.

With ISO-certified labs and expertise in global sample handling, we deliver fast turnaround (10–14 days) across platforms: Illumina , DNBseq, and Salus.

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Labema's wide range of diagnostical products includes thousands of products from approximately 50 different manufacturers from all over the world. In our genetic portfolio we are working with well-known brands such as SOPHiA Genetics and Hamilton Robotics.

Our mission is to provide state-of-the-art solutions that enable our customers to make the world healthier, cleaner and safer. We have been dedicated to providing quality medical and laboratory equipment for 27 years. Whether you are a startup lab or an experienced practitioner, we are here to support you every step of the way. In addition, we ensure the smooth operation of your laboratory thanks to our top-notch technical support team

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Nova Natura is a local sales and service partner for instruments, reagents and consumables used in laboratories and biopharmaceutical industry processes in Estonia, Latvia and Lithuania. Our team of product, application and service specialists, experienced in different fields of life sciences, are keen to work together with local users for choosing the solutions that fit best the users needs

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Surgitech, founded in 1998, is an Estonian company and the preferred supplier for medical institutions and research laboratories across the country. Our product range upholds top-quality standards and supports both clinical diagnostics and scientific research. At GeneForum 2025, stop by our booth to see and try the latest instruments, watch live demos and chat with our team of experts!

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Triolab is an expert company that sells diagnostics and bioresearch products in Finland, Estonia, Latvia, and Lithuania. Triolab provides com-prehensive product and technical support for the products it represents. Triolab´s range of products consists of solutions that use modern meth-ods for the diagnostics of genetic illnesses and susceptibility, as well as cancer mutations, pharmacogenetics and chromosome abnormalities. The range includes both rapid and user-friendly tests conducted directly from sample material, advanced next-generation sequencing (NGS) tests, Optical Genome Mapping (OGM) and quality assurance products.

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