DNA Variation Matters

Past, Present & Future of Human Genomics

Why this symposium?

The analysis of DNA variations, including Single Nucleotide Polymorphisms (SNPs), is a standard research approach to understand causes of disease, in particular the so-called "complex" diseases such as diabetes, osteoporosis, cancer, Alzheimer disease, etc. Since the Human Genome project, the field has been changing fast with large scale projects, biobanks, and databases (e.g., UK Biobank, TopMed, GWAS catalogues), and novel technology being continuously introduced, including Next Generation Sequencing, microarrays, single cell technologies, iPS, and multi-omics. In the past decade we have also witnessed a slow but steady movement of these discoveries and potential applications into clinical and public health settings with substantial progress being made in pharmacogenetics and polygenic risk scores. In addition, the use of genetic information has also found applications beyond healthcare, in areas such as archeological genetics and forensic genetics, making it an increasingly relevant topic in the public debate.

This symposium brings together a star-studded cast of international top-experts in diverse areas in this exiting and fast developing field to give us *state-of-the-art* insights where we stand and what the future might bring. The symposium also celebrates the 20th anniversary of the infamous SNP course organized by the Laboratory for Population Genomics at Erasmus MC. In this course several of the speakers have featured as either participant and/or speaker, and it has trained several hundreds of students and interested scientists over the years in this area. The symposium forms the closing two days of the regular SNP course program for which further information will follow in due time.

Audience and participation

The symposium is organized for all academic levels and for all scientific backgrounds, but is somewhat advanced and requires some background knowledge of the central molecular biological dogma's (e.g., DNA- and gene-structure, DNA encodes RNA encodes protein, etc.) and of basic genetic principles. We welcome professors, senior scientist, PhD students, postdocs, and technicians, clinicians as well as basic scientists, and from all scientific disciplines (from medicine to biology, and epidemiology, psychology to economics). The symposium is open to all scientific institutes in- and outside the Netherlands, such as the Erasmus MC Postgraduate School Molecular Medicine, NIHES, and MGC, and other research schools, as well as for other universities in- and outside the Netherlands.

Erasmus MC
University Medical Center Rotterdam

Program Thursday 23 November 2023

Queridozaal + Foyer Q

Session 1: (Complex) Genetics in the Clinic; chair: Linda Broer

Time	*Speaker*	*Subject*
9.15-9.50	***Walk in & coffee***	
9.50-10.00	**André Uitterlinden: Opening Remarks**	
10.00-10.30	**Veronika Lipphardt** University College Freiburg, Germany	**Genetic research of isolated populations from the viewpoint of social studies of science**
10.30-11.00	**Jesse Swen** Leiden University Medical Center, Leiden, NL	**Pharmacogenetics – from promise to practice**
11.00-11.30	***Break***	
11.30-12.00	**Rens Reeskamp** Amsterdam UMC, NL	**Genetic testing and polygenic scores in Familial Hypercholesterolemia**
12.00-12.30	**Jeroen van Rooij** Erasmus MC, Rotterdam, NL	**The Genotyping On ALL patients (GOALL) project**
12.30-13.00	**Bartha Knoppers** McGill University, Canada	**Ethico-Legal "Futures" in Genomic Medicine**

Lunch and time for networking: 13:00-14:00h

Session 2: Big, Bigger & Biobanks in Genetics; chair: Fernando Rivadeneira

Time	*Speaker*	*Subject*
14.00-14.30	**Michael Cook** Our Future Health, UK	**Our Future Health – a new translational prospective cohort study**
14.30-15.00	**Segun Fatumo** MRC/UVRI Uganda and London School of Hygiene and Tropical Medicine, London, UK	**African biobanks: the most genetic diverse continent explored**
15.00-15.30	**Andres Metspalu** University of Tartu, Estonia	**Estonian Biobank: the Estonian Population**
15.30-16.00	***Break***	
16.00-16.30	**Samuli Ripatti** University of Helsinki, Finland	**Context Matters for Genetic Risk**
16.30-17.00	**Matt Brown** Genomics England, UK	**Genetics into the clinic and back. The Genomics England experience**

Keynote Lecture (17.00-17.45):

Manfred Kayser

Erasmus MC, Rotterdam, the Netherlands

Genetic Identity

18.00-19.00h : drinks

19.30h : group dinner Tai Wu restaurant, Rotterdam

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Program Friday 24 November 2023

OWR36 + Foyer C

Session 3: From Genetics to Causal Biology; chair: Cindy G. Boer

Time	*Speaker*	*Subject*
10.00-10.30	**Stefan Barakat** Erasmus MC, Rotterdam, NL	**Finding causes of missing heritability in neurogenetic disorders: exploring the dark matter of the genome**
10.30-11.00	**Musa Mhlanga** Radboud University, Nijmegen, NL	**RNA Epigenetics an emergent property of DNA variation**
11.00-11.30	***Break***	
11.30-12.00	**Struan Grant** University of Pennsylvania / Children's Hospital of Philadelphia, USA	**Variant-to-gene mapping for common complex traits**
12.00-12.30	**Lude Franke** University Medical Center Groningen, Groningen, NL	**Multi-omics approaches to understand disease**
12.30-13.00	**Karol Estrada** VP Translational Genomics, Maze Therapeutics, Boston, USA	**Designing Therapies with Nature's Codebook: Human Genetics**

Lunch and time for networking: 13:00-14:00

Session 4: Complex Genetics and disease; chair: André Uitterlinden

Time	*Speaker*	*Subject*
14.00-14.30	**Doug Easton** University of Cambridge, UK	**Breast Cancer genetics in diagnostics and prevention**
14.30-15.00	**Stuart Ralston** University of Edinburgh, UK	**Genetics to help diagnostics and treatment of Paget's disease**
15.00-15.30	**Felix Day** University of Cambridge, UK	**Reproductive genetics in health and disease**
15.30-16.00	***Break***	
16.00-16.30	**Inga Prokopenko** University of Surrey, UK	**From genetics of glycaemic health to diabetes complications and comorbidities**
16.30-17.00	**Eleftheria Zeggini** Helmholtz Institute of Translational Genomics, Technische Universitat Munich, Klinikum rechts der Isar, Munich, Germany	**Translational Genomics of Osteoarthritis**

17.00-19.00h : networking drinks

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