



GENETICS RETREAT

ANNUAL MEETING #30



Programme

Pre-Retreat & Genetics Retreat



Maastricht UMC+

Pre-Retreat workshop: Your career - what's next?

Wednesday 3 May 2023

ROOM 1

- 13.30 - 13.40** Registration and welcome with coffee/tea
- 13.40 - 13.45** Opening
- 13.45 - 14.15** Preparing yourself for the next step
(post-doctorate) in your career
John Creemers (KU Leuven)
- 14.15 - 14.45** Women in science; novel Recognition & Awards
Berta Cillero Pastor (Maastricht University)
- 14.45 - 15.15** From Rolduc to Medical Scientific Liaison
Lara Hochfeld (University of Bonn, Gilead Sciences)
- 15.15 - 15.35** Coffee break
- 15.35 - 16.05** European post-doc funding; calls/strategies/
planning
Rachel van Swelm (Radboudumc Nijmegen,
Grant Office)
- 16.05 - 16.35** My best career advise & closure Pre-Retreat
Alex Hoischen, Kerstin Ludwig, Willem Voncken
- 16.35 - 17.05** Enabling personalized medicine
Patrick Short (Sano Genetics)
- 17.05 - 17.30** Open discussion and Q&A
All participants & presenters

GROTE EETZAAL

- 17.30 - 19.00** Dinner

Genetics Retreat

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Thursday 4 May 2023

ROOM 2

- 10.30 - 11.00** Registration and collect name badge
Welcome with Limburgse vlaai

ROOM 1

- 11.00 - 11.10** Opening
Willem Voncken
- 11.15 - 11.30** Population Genetics
A genetic analysis of familial aggregation in inflammatory bowel disease multiplex families
Deborah Jans (Leuven)
- 11.30 - 11.45** Prevalence and Disease Expression of Pathogenic and Likely Pathogenic Variants Associated With Inherited Cardiomyopathies in the General Population
Marion van Vugt (Utrecht)
- 11.45 - 12.00** The search for longevity genes in the Dutch population with the Longevity Families of the Netherlands study (LOF-NL)
Pasquale Putter (Leiden)

12.00 - 12.15 **Polygenic score analyses of subjective well-being in the German TwinLife sample**
Charlotte Pahnke (Bonn)

12.15 - 12.30 *Integrated session discussion*

ROOM 2

12.30 - 13.30 **Lunch**

ROOM 1

Reproductive Genetics

13.30 - 13.15 **The power of parent driven research – 10 years Chromosome 6 Project**
Aafke Engwerda (Groningen)

13.15 - 13.30 **Simulated reproductive carrier screening in the perinatal genomic autopsy study**
Lauren Akesson (Nijmegen)

13.30 - 13.45 **Using low-coverage non-invasive prenatal testing data for polygenic risk score calculation**
Sara Becelaere (Leuven)

13.45 - 14.00 **The nature and prevalence of chromosomal dynamics in first trimester spontaneous pregnancy loss**
Rick Essers (Maastricht)

14.00 - 14.15 **Clinical whole-genome sequencing-based haplarithmisis enables simple, scalable and universal preim-plantation genetic testing**
Anouk Janssen (Maastricht)

14.15 - 14.30 *Integrated session discussion*

ROOM 2

14.30 - 15.00 **Coffee break**

ROOM 1

Genotypes, Phenotypes & Environments

15.00 - 15.15 **Studying Microbiome-dependent Ageing With Germ-Free Mice: A Mouse Ageing Atlas**
Ward Nijen Twilhaar

15.15 - 15.30 **Methylome-wide association studies in major psychiatric disorders**
Friederike David (Bonn)

15.30 - 15.45 *Integrated session discussion*

15.45 - 15.55 **From genes to proteoforms to explore disease biology**
Lars Vahlkamp (Seer) on behalf of life & brain

16.00 - 17.30 **Jubilee programme of your choice or free time**

GROTE EETZAAL

18.00 - 19.30 **Dinner**

ROOM 1

19.30 - 20.30 **New approaches to cancer therapy: if you do what you did, you get what you got**
René Bernardts (NKI, Amsterdam, Leiden)

At 20.00 hours two minutes of silence to commemorate the victims of the Second World War in Europe and Southeast Asia and the victims of war situations and peacekeeping operations in which the Netherlands were involved after the Second World War.

ARCHED CELLAR VERLOREN ZOON

20.30 - 23.30 **Social Evening with a jubilee surprise**

Friday 5 May 2023

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07.00 - 09.00 **Breakfast + checking out at the reception desk**

ROOM 1

Medical Genetics

09.15 - 09.20 **Opening**
Kerstin Ludwig

- 09.20 - 09.30** **Genome Services Maastricht UMC+**
Wanwisa van Dijk
- 09.30 - 09.45** **Comprehensive de novo mutation discovery with HiFi long-read sequencing**
Bart van der Sanden (Nijmegen)
- 09.45 - 10.00** **Generating human models to study and treat lysine metabolism disorders**
Imke Schuurmans (Nijmegen)
- 10.00 - 10.15** **Exome reanalysis in a cohort of patients with inborn errors of immunity**
Emil Vorsteveld (Nijmegen)
- 10.15 - 10.25** ***Integrated session discussion***

CHURCH

- 10.30 - 11.00** **Jubilee break: organ concert**
Tjeu Zeijen

ROOM 1

- 11.00 - 11.15** **Coffee break**
- Molecular Genetics & Mechanisms**
- 11.15 - 11.30** **The impact of loss-of-function genes on lipid and lipoprotein biomarkers in UK Biobank**
Abdulrahman Alasiri (Utrecht)
- 11.30 - 11.45** **Allele-specific expression analysis for complex genetic phenotypes applied to a unique dilated cardiomyopathy cohort**
Daan van Beek (Maastricht)
- 11.45 - 12.00** **Targeted enrichment of the OTOA gene through the use of the Samplix Xdrop® Sort**
Nick Zomer (Nijmegen)
- 12.00 - 12.15** **Curation and expansion of Human Phenotype Ontology for Systemic Auto inflammatory Disease SAID improves phenotype-driven disease matching**
Kars Maassen (Groningen)
- 12.15 - 12.30** ***Integrated session discussion***

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- 12.30 - 13.30** **Lunch**
- 13.30 - 14.00** **Award ceremony & photo shoot prize winners and all participants**
Alexander Hoischen *on behalf of Pacific Biosciences*