

23-24 Apr 2009 60 jarig jubileum NVHG

Genetics into the Future

NVHG voorjaarsvergadering - Nederlandse Vereniging voor Humane Genetica (www.nvhg-nav.nl)

60-jarig jubileum NVHG , 2- daagse

Golden Tulip Conference Hotel Koningshof, Veldhoven

Program Thursday April 23

10.00-10.30 Registration (untill 11.00 hrs. registration open)

10.30-12.30 Plenary session

Auditorium

Chair: R. Hofstra

10.30-10.45 Opening: R. Hofstra

10.45-11.15 **Martinus Niermeyer** (UMC St Radboud, Nijmegen)

"On 60 years of genes and genetics in the Delta" (1)

11.15-11.45 **Hanne Meijers** (VUMC, Amsterdam)

The future of Clinical Genetics (2)

11.55-12.35 **Nigel Carter** (Sanger Institute, Cambridge)

The future of Cytogenetics (3)

12.35-14.00 Lunch, huishoudelijke vergadering(en)

14.00 -15.10 **Parallel sessions**

14.00-15.10 **Symposium 1A: Clinical Genetics**

Auditorium

Chair: Joke Verheij

14.00-14.25 **Suzanne Frints** (MUMC, Maastricht)

Beauty and The Beast: About X-linked Mental Retardation Disorders and How X-Dosage Regulation Can Contribute to The Clinical Phenotype in Females (4)

14.25-14.40 **Mike Gerards** (MUMC, Maastricht)

Homozygosity mapping reveals new genes and gene defects in consanguineous families with OXPHOS disease (5)

14.40-14.55 **Birgit Sikkema-Raddatz** (UMCG, Groningen)

Nine patients with deletion 15q11.2 between breakpoints 1 and 2 of the prader-willi/ angelman critical region; clinically relevant or not? (6)

14.55-15.10 **Bregje van Bon** (UMCN, Nijmegen)
Further delineation of the 15q13.3 microdeletion and duplication syndromes: A clinical spectrum varying from non-pathogenic to a severe outcome (7)

14.00-15.15 **Symposium 1B: Novel developments**

Parkzaal

Chair: Lies Hoefsloot

14.00-14.15 **Elmar Tobi** (LUMC, Leiden) 15 minutes

Epigenetic plasticity and prenatal exposure to famine (8)

14.15-14.25 **Maartje Vogel** (UMCU, Utrecht) 10 minutes

Implementation of Roche 454 sequencing technology in a diagnostic setting (9)

14.25-14.35 **Hans Scheffer** (UMCN, Nijmegen) 10 minutes

Parallel sequencing of multiple genes in genetically heterogeneous disorders after array-based enrichment (10)

14.35-14.45 **Astrid Out** (LUMC, Leiden) 10 minutes

Estimation of MUTYH variant frequencies in pooled DNA with massive parallel sequencing (11)

14.45-15.00 **Annemieke Aartsma-Rus** (LUMC, Leiden) 15 minutes

First clinical trial on therapeutic antisense-mediated exon skipping for Duchenne muscular dystrophy (12)

15.00-15.15 **Femke de Vrij** (Erasmus MC, Rotterdam) 15 minutes

Rescue of behavioral phenotype and neuronal protrusion morphology in Fmr1 KO mice (13)

15.10-17.00 Posters, coffee, tea

Presenters at posters

17.00 **Lodewijk Sandkuijl lecture**

Auditorium

Chair: Gerard te Meerman

Paul de Bakker (Broad Institute, Boston)

What genome-wide association studies have taught us about human disease (14)

Drinks

Dinner

Feest/Party

Program Friday April 24

- 9.00-10.00 **Plenary session:**
Auditorium
Chair: Gert Jan van Ommen
- 9.00-9.30 **Joep Geraedts** (MUMC, Maastricht)
PGD and its future (15)
- 9.30-10.00 **Robert Passier** (LUMC, Leiden)
Human embryonic stem cells for cardiac repair and drug discovery (16)
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10.05 -11.15 Parallel sessions

- 10.05-11.15 **Symposium 2A: Neurogenetics**
Parkzaal
Chair: Frank Baas
- 10.05-10.30 **Gert Scheper** (VUMC, Amsterdam)
Mutations in *DARS2*, encoding mitochondrial aspartyl-tRNA synthetase, are the cause of the childhood white matter disorder LBSL (17)
- 10.30-10.45 **Jessica de Greef** (LUMC, Leiden)
Contraction-dependent (FSD1) and independent (FSD2) epigenetic changes of D4Z4 underlie FSD (18)
- 10.45-11.00 **Yunia Sribudiani** (UMCG, Groningen)
Functional Studies on Ret Non-Coding Region Mutations and The Involvement of the Nxf/Arnt2; Sim2/Arnt2 Heterodimers in The Regulation of RET Expression (19)
- 11.00-11.15 **Maria Alves** (UMCG, Groningen)
A functional approach to understand the function of the Kinesin Binding Protein (KBP) in Goldberg-Shprintzen syndrome (20)
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10.05 -11.15 Symposium 2B: Oncogenetics

- Auditorium
Chair: Marjolijn Ligtenberg
- 10.05-10.30 **Ron Smits** (Erasmus MC, Rotterdam)
Selection for somatic mutations within the β -catenin signalling pathway: Can it explain the localization of colorectal tumors? (21)
- 10.30-10.45 **Marjolijn Ligtenberg** (UMCN, Nijmegen)
A novel genetic mechanism for Lynch syndrome: heritable somatic methylation and inactivation of MSH2 due to deletion of the 3' exons of the upstream gene (22)
- 10.45-11.00 **Gerben Duns** (UMCG, Groningen)
Differentially expressed microRNAs in clear cell renal cell carcinoma (23)

11.00-11.15 **Eva Wielders** (NKI, Amsterdam)
Functional characterization of unclassified MSH2/MSH6 variants found in patients with (suspected) HNPCC using oligonucleotide-directed mutagenesis (24)

11.15-11.45 Coffee-tea break

11.45-12.55 Parallel sessions

11.45-12.55 Symposium 3A: Mechanisms

Auditorium

Chair: Bert de Vries

11.45-12.10 **Maurice van den Hoff** (AMC, Amsterdam)
Separation of the myocardial- and epicardial-lineages at the inflow of the forming heart is regulated by an interaction between BMP and FGF signaling (25)

12.10-12.25 **Elisabeth Lodder** (Erasmus MC, Rotterdam)
Co-localization of the ZRS with Sonic Hedgehog in the developing limb bud (26)

12.25-12.40 **Lisenka Vissers** (UMCN, Nijmegen)
Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture (27)

12.40-12.55 **Hans van Bokhoven** (UMCN, Nijmegen)
Origins of mental retardation across species: Clinical, molecular and functional characterization of Euchromatin Histone Methyltransferase 1 mutations (28)

11.45-12.55 Symposium 3B: Complex Genetics

Parkzaal

Chair: Peter Heutink

11.45-12.10 **Lude Franke** (UMCG, Groningen)
What is wise in the post-GWA era: Chasing the (rare) causal variants and/or functional follow up? (29)

12.10-12.25 **Martin Poot** (UMCU, Utrecht)
Novel loci and candidate genes for autism spectrum disorder detected by SNP array based segmental aneuploidy screening (30)

12.25-12.40 **Sasha Zhernakova** (UMCU, Utrecht)
From shared genetics to shared pathogenesis of immune-related diseases (31)

12.40-12.55 **Yurii Aulchenko** (Erasmus MC, Rotterdam)
Genome-wide association analysis of 16 European populations identifies novel loci influencing lipid levels (32)

12.55 -13.55 Lunch and posterviewing

13.55-14.10 Auditorium: **Uitreiking Young Scientist Award 2009**
Uitreiking Young Scientist Travel Award 2009

Chair: Frank Baas & Robert Hofstra

14.10-15.55 **Plenary session**

Auditorium

Chair: Robert Hofstra

14.10-14.40 **Han Brunner** (UMC St Radboud, Nijmegen)
Monogenic diseases: The end of the beginning (33)

14.40-15.10 **Cisca Wijmenga** (UMCG, Groningen)
Towards unraveling the genetic puzzle of immune-related disease (34)

15.10-15.55 **Gert Jan van Ommen** (LUMC, Leiden)
Human Genetics – from genes to therapies (35)

15.55 Sluiting
chair: Robert Hofstra