nederlandse vereniging voor
Humane Genetica

September 20 & 21, 2012
Putting next generation sequencing in perspective

The NVHG Two-Day Autumn Symposium 2012 - Dutch Society of Human Genetics
Nederlandse Vereniging voor Humane Genetica (www.nvhg-nav.nl)

Program Thursday September 20, 2012

10:00-10:30 Registration (open until 11.00 hrs)

10.30-12.25 **Plenary session**
Room: Athene B/C
Chair: Robert Hofstra

10.30-10.40 Opening: Robert Hofstra

10.40-11.15 **Dorret Boomsma** (VU Amsterdam)
Twins in the NGS era (1)

11.15-11:50 **Eline Slagboom** (LUMC Leiden)
NGS for age-related disease and longevity, how to make the genome confess (2)

11:50-12.25 **Edwin Cuppen** (UMC Utrecht)
From genetic inventories of congenital patients towards molecular understanding of phenotypes (3)

12.30-14.00 Lunch

13:00-14:00 Huishoudelijke vergadering VKGN
Room: zaal 6

13.15-14:00 Huishoudelijke vergadering VKGL
Room: zaal 7

14.00-16.00 **Parallel sessions**

14.00-16.00 **Symposium 1A: VKGN "Syndromes & exomes"**
Room: Athene A
Chair: Alice Brooks

14.00-14.20 **Ilse Feenstra** (UMC ST Radboud Nijmegen)
Whole exome sequencing in a diagnostic setting; experiences of clinical geneticists and patients (4)

14.20-14.40 **Jeske van Harssel** (UMC Utrecht)
Dominant missense mutations in ABCC9 cause Cantú syndrome (5)

14.40-15.00 **Gijs Santen** (LUMC Leiden)
Where phenotype meets genotype: Coffin-Siris syndrome as an example (6)

15.00-15.20 **Sietske Kevelam** (VU Amsterdam)
Exome sequencing identifies SLC19A3 mutations in young infants with an acute dramatic, encephalopathy (7)

15.20-15.40 **Suzanne Sallevelt** (MUMC Maastricht)
Next-generation sequencing (NGS) in mitochondrial diseases (-)

15.40-16.00 **Bianca van den Bosch** (MUMC Maastricht)
Exome sequencing reveals a frequent Moroccan founder mutation in SLC19A3 as a new cause of early-childhood fatal Leigh syndrome (8)

14.00-16.00 **Symposium 1B: VKGL**
Room: Athene B/C
Chair: Bert Bakker & Hans Scheffer

14.00-14.30 **Luc Dehaspe** (Universiteit Leuven, België)
Quality aspects of diagnostic NGS (9)

14.30-15.00 **Elles Boon** (LUMC Leiden) & **Gerard te Meerman** (UMC Groningen)
NGS & NIPD: the NITRO study (10 / 11)

15.00-15.20 **Lisenka Vissers** (UMC St. Radboud Nijmegen)
Diagnostic Exome Sequencing in Patients with Severe Intellectual Disability (12)

15.20-15.40 **Terry Vrijenhoek** (UMC Utrecht)
Beyond NGS technology – overcoming key challenges for nation-wide implementation of next-generation sequencing into routine diagnostics (13)

15.40-16.00 **Steven Van Vooren** (Cartagenia, Leuven, België)
Clinical NGS data analysis with Cartagenia BENCH lab NGS: Store, Explore, Automate and Validate. (14)

14.00-16.00 **Symposium 1C: NACGG**
Next-generation sequencing: return of incidental findings in biobanking and clinical practice

Room: zaal 6
Chair(s): Cecile Janssens + Irene van Langen

14.00-14.20 **Helger IJntema** (UMC St. Radboud Nijmegen)
Incidental findings from exome sequencing: what can we tell from our DNA?

14.20-14.40 **Ellen Smets** (AMC Amsterdam)
Always expect the unexpected: preferences of donors and researchers for the return of results in biobank research

14.40-15.00 **Tessel Rigter** (VU Amsterdam)
Next generation sequencing in diagnostics: exploring needs for the informed consent procedure

15.00-15.20 **Annelien Bredenoord** (UMC Utrecht)
Feedback of results: a menu of options

15.20-15.40 Discussion

16.00-17.00 Posters, coffee, tea
Presenters at posters

17.00-18.30 **Forum discussion**
in Dutch
Room: Athene B/C

Prof. dr. Bert Schadé
Emeritus hoogleraar huisartsgeneeskunde

Prof. dr. Dick Willems
Hoogleraar Medische Ethiek, afdeling huisartsgeneeskunde AMC / UVA,
Lid Raad voor Gezondheid

Prof. mr. Simon Stolwijk
Emeritus hoogleraar strafrecht

Marjolein Bos
Nierpatiënten Vereniging Nederland (NVN) - Kennisgroep Erfelijke Nierziekten

Evening **Drinks** **Dinner** **Party**

Program Friday September 21, 2012

9.00-10.00 **Plenary session**
Room: Athene B/C
Chair: Hans Kristian Ploos van Amstel

9.00-9.30 **André Uitterlinden** (Erasmus MC Rotterdam)
Genome-Wide Explorations in Cohort Studies (20)

9.30-10.00 **Hans Scheffer** (UMC St. Radboud Nijmegen)
Development, validation and implementation of next generation sequencing in clinical genetic diagnostics (21)

10.15-11.15 **Parallel sessions**

10.15-11.15 **Symposium 2A: Gene Discovery**
Room: Athene B/C
Chair: Raoul Hennekam

10.15-10.30 **Richarda de Voer** (UMC St. Radboud Nijmegen)
Mutations in the Spindle Assembly Checkpoint Genes BUB1 and BUB3 Are Risk Factors for Mosaic Aneuploidy and Colorectal Cancer (22)

10.30-10.45 **Magdalena Harakalova** (UMC Utrecht)
X-exome sequencing identifies a HDAC8 variant in a large pedigree with X-linked intellectual disability, truncal obesity, gynecomastia, hypogonadism and unusual face (23)

10.45-11.00 **Richard Lemmers** (LUMC Leiden)
Identification of the epigenetic modifier of the D4Z4 epiallele in FSHD type 2 (24)

11.00-11.15 **Tjitske Kleefstra** (UMC St. Radboud Nijmegen)
Disruption of an EHMT1-associated chromatin modification module causes intellectual disability (25)

10.15-11.15 **Symposium 2B: Pharmacogenetics**
Room: Athene A
Chair: Peter Devilee

10.15-10.30 **Marieke Coenen** (UMC St. Radboud Nijmegen)
Screening of the TPMT gene before thiopurine treatment results in a lower leucopenia occurrence in patient with inflammatory bowel disease (26)

10.30-10.45 **Martina Wilke** (Erasmus MC Rotterdam)
Assessment of CFTR function in patients with CFTR mutations of unknown clinical relevance (27)

10:45-11:00 **Elmar Tobi** (LUMC Leiden)
Genome-scale characterization of DNA methylation differences after prenatal exposure to the Dutch Famine using next generation bisulfite sequencing (28)

11.00-11.15 **Catherina Meijer** (Erasmus MC Rotterdam)
Is Direct-to-Consumer Personal Genome Testing for All Ethnicities? (29)

11.15-11.45 Coffee-tea break

11.45-12.45 **Parallel sessions**

11.45-12.45 **Symposium 3A: Gene function & Brain**
Room: Athene A
Chair: Hans van Bokhoven

11.45-12.00 **Grazia Mancini** (Erasmus MC Rotterdam)
Mutations in Rotatin link primary cilia function to organization of the human cerebral cortex (30)

12.00-12.15 **Renate Hukema** (Erasmus MC Rotterdam)
A new inducible mouse model for Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) (31)

12.15-12.30 **Gea Beunders** (VU Amsterdam)
C-terminal deletions of the AUTS2 locus cause distinct syndromic features and cognitive impairment (32)

12.30.12.45 **Erik-Jan Kamsteeg** (UMC St. Radboud Nijmegen)
Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of alpha-dystroglycan (33)

11.45-12.45 **Symposium 3B: Diagnostics**
Room: Athene B/C
Chair: Lies Hoefsloot

11.45-12.00 **Christian Gilissen** (UMC St. Radboud Nijmegen)
Exome sequencing analysis for diagnostics (34)

12.00-12.15 **Isaaec Nijman** (UMC Utrecht)
Bioinformatic processing of next generation sequencing data in a diagnostic setting (35)

12.15-12.30 **Arthur van den Wijngaard** (MUMC Maastricht)
CHIP-based parallel sequencing of multiple candidate genes resolves the genetic architecture of patients with cardiomyopathy (36)

12.30-12.45 **Rowina Almomani** (UMC Groningen)
Exome sequencing and haplotype sharing analyses result in the identification of novel genetic causes of cardiomyopathies (37)

12.45-13.00 Algemene ledenvergadering NVHG

13:00-14:00 Lunch and posterviewing

14.00-15.45 **Plenary session**
Room: Athene B/C
Chair: Frank Baas

14.00-14.35 **Rene Bernards** (AVL Amsterdam)
Using functional genetics to optimize the treatment of cancer (38)

14.35-15.10 **Henk Stunnenberg** (NCMLS, UMC St. Radboud Nijmegen)
Human epigenomes: the good, the bad and the ugly (39)

15.10-15.45 NVHG Annual Award 2012

15.15-16.00 Prof. dr. Hans Galjaard Lecture 2012

Gert Matthijs (Universiteit Leuven, België) (40)
Next Generation Sequencing and Diagnostics: the Sky is the limit but this place is Earth

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16:00 **Closure**