

# PROGRAMME GENETICS RETREAT NVHG GRADUATE MEETING



**THURSDAY 17 MAY 2018**

10.00 - 11.00	Registration & collect name badge <b>Welcome with Limburgse vlaai</b>	<b>Room 3</b> <b>Foyer &amp; Room 4</b>
11.00 - 11.15	Opening by Willem Voncken	<b>Room 2</b>

## COMPLEX GENETICS Room 2

11.15 - 11.30	Nina Ishorst, <i>Bonn</i>	Large-scale resequencing study of nsCL/P candidate genes in 1061 nsCL/P cases and 1591 controls
11.30 - 11.45	Doris Skoric-Milosavljevic, <i>Amsterdam</i>	Genome-wide association study of transposition of the great arteries
11.45 - 12.00	Frederic Thieme, <i>Bonn</i>	Targeted resequencing of non-coding risk loci for complex traits using single-molecule molecular inversion probes (smMIPs)
12.00 - 12.15	Eva van Walree, <i>Amsterdam</i>	Genetic homogeneity in DARWIN, an isolated population in the Netherlands
12.15 - 12.25	Discussion & Closure	

12.25 - 13.30 **Lunch** **Foyer & Room 4**

## CANCER GENETICS Room 2

13.30 - 13.45	Evelien Hurkmans, <i>Nijmegen</i>	Pharmacogenetics of chemotherapy response in osteosarcoma: a genetic variant in SLC7A8 is associated with progressive disease
13.45 - 14.00	Natasha Van Poppelen, <i>Rotterdam</i>	Are iris melanomas a different subgroup in uveal melanomas based on their genetic makeup?
14.00 - 14.15	Julia Schröder, <i>Bonn</i>	Barrett's esophagus and esophageal adenocarcinoma: systematic integration of eQTL data and candidate loci association study
14.15 - 14.30	Alexej Knaus, <i>Bonn</i>	An increased rate of multisite de novo mutations in offsprings of fathers exposed to high doses of irradiation
14.30 - 14.45	Mohamed Alimohamed, <i>Groningen</i>	DNA test for translocation detection in acute leukemia using targeted locus amplification
14.45 - 14.55	Discussion & Closure	

14.55 - 15.30 **Coffee break & Pitch by GC Biotech** **Foyer & Room 4**

## STRUCTURAL VARIATION & FUNCTIONAL GENOMICS Room 2

15.30 - 15.45	Masoud Zamani Esteki, <i>Maastricht</i>	Human in vitro fertilization has no effect on prevalence of mosaic copy-number alterations in fetal and placental lineages
15.45 - 16.00	Miaozhen Huang, <i>Groningen</i>	Identification of modifiers of the age of onset variance and disease progression in a Dutch cohort of Machado-Joseph disease patients
16.00 - 16.15	Judith Vlaar, <i>Utrecht</i>	Detection of structural variants and understanding the consequences on the molecular mechanisms of the genome
16.15 - 16.30	Carola Greve, <i>Bonn</i>	Exome-wide copy number variant analysis in multiply affected families provides new insight into the pathogenesis of schizophrenia
16.30 - 16.45	Julia Welzenbach, <i>Bonn</i>	Integration of large-scale genetic and functional data sets enable mechanistic insights into craniofacial development and disease
16.45 - 16.55	Discussion & Closure	

16.55 - 17.10 **Power break & Pitch by Eppendorf** **Foyer & Room 4**

## SCIENCE IN OPEN SPACE Foyer & Room 4

17.10 - 18.15	Open Space Team discussions	Choose a table/team and discuss the topics which all of you have chosen
18.15 - 18.35	Open Space Team Pitches	over an aperitif

18.35 - 20.00 **Walking dinner** **Foyer & Room 4**

## NVHG KEYNOTE LECTURE Room 2

20.00 - 20.45	Cisca Wijmenga, <i>Groningen</i>	We and our small friends: ins and outs of gut microbiome
20.45 - 21.00	Discussion with keynote speaker	

21.00 - 23.00 **Social Evening** **Boerenkelder**

# PROGRAMME GENETICS RETREAT NVHG GRADUATE MEETING



**FRIDAY 18 MAY 2018**

07.00 - 09.00 **Breakfast**

**Grote Eetzaal**

## MODELS AND TECHNOLOGY

**Room 2**

09.00 - 09.15 Michaela Bartusel, *Köln*

Molecular and functional characterization of novel loci associated with orofacial clefting

09.15 - 09.30 Christa de Geus, *Groningen*

Investigating the cerebellum in CHARGE syndrome: a conditional cerebellar knockout model of Chd7

09.30 - 09.45 Helen Roessler, *Utrecht*

Efficient CrispR/Cas9-based nucleotide editing to model cardiovascular anomalies of Cantú syndrome in zebrafish

09.45 - 10.00 Bart Appelhof, *Leiden*

A Zebrafish Model for Pontocerebellar Hypoplasia

10.00 - 10.15 Iliana Michailidou, *Leiden*

Systemic inhibition of the membrane attack complex impedes chronic neuroinflammation

10.15 - 10.25 Discussion & Closure

10.25 - 11.00 **Coffee break**

**Foyer & Room 4**

## NOVEL GENES; RARE DISORDERS

**Room 2**

11.00 - 11.15 Marjoleine Broekema, *Utrecht*

Deciphering the molecular defect of a single complex AGPAT2 allele in the spectrum of genetic lipodystrophies

11.15 - 11.30 Gabriel Dworschak, *Bonn*

Mutations in SHROOM4 in VATER/VACTERL association and characterization in a zebrafish model

11.30 - 11.45 Alina Hilger, *Bonn*

Mutations in BNC2 Lead to Autosomal-Dominant Lower Urinary Tract Obstruction (LUTO)

11.45 - 12.00 Evelien Slot, *Rotterdam*

Genetic testing of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins

12.00 - 12.15 Tessa van Dijk, *Leiden*

Mutations in COASY cause prenatal onset microcephaly, pontocerebellar hypoplasia and arthrogryposis

12.15 - 12.25 Discussion & Closure

12.25 - 13.30 **Lunch**

**Grote Eetzaal**

13.30 - 13.40 **Award ceremony by Alexander Hoischen**

**Room 2**

13.40 - 13.50 **Photo shoot prize winners & all participants**

**Enclosed garden**