PROGRAMME GENETICS RETREAT
NVHG GRADUATE MEETING

GENETICS ROLDUC MEETING RETREAT

THURSDAY 28 MARCH 2019

10.00 - 10.30	Registration and collect name badge		**Room 3**
	Welcome with Limburgse vlaai		**Foyer & Room 4**
10.30 - 10.45	Opening by Willem Voncken		**Room 2**

FRONTLINE GENETICS — Room 2

10.45 - 11.00	Carina Mathey (University Hospital Bonn)	Design of a candidate smMIPS sequencing study in patients with hereditary angioedema of unknown cause (U-HAE) with further application in patients with ACE-inhibitor induced angioedema
11.00 - 11.15	Eline Simons (University of Antwerp)	Study of the contribution of SCN10A mutations to the Brugada syndrome genetic architecture
11.15 - 11.30	Jolijn Verseput (Radboudumc Nijmegen)	Clustering missense mutations in WDR5 cause a new neurodevelopmental disorder
11.30 - 11.45	Sophia Schneider (University Hospital Bonn)	Genetic drivers of congenital Hydro- and Chylothoraces
11.45 - 12.00	Amber de Haan (UMC Groningen)	Validation of diagnostic algorithms and identification of novel genes in young patients with unknown primary renal disease (VARIETY)
12.00 - 12.15	Jeroen Smits (Radboudumc Nijmegen)	De novo and inherited loss-of-function variants of ATP2B2 are associated with rapidly progressive hearing impairment
12.15 - 12.25	Discussion and closure	

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

PRE-CLINICAL ANIMAL MODELS — Room 2

13.30 - 13.45	Carmen Daems (KU Leuven)	A20: a predisposing factor for neuropsychiatric lupus
13.45 - 14.00	Helen Roessler (UMC Utrecht)	Loss-of-function mutation of ABCC9 causes ABCC9-related Intellectual disability Myopathy Syndrome (AIMS) – a novel KATP channelopathy
14.00 - 14.15	Joost Kummeling (Radboudumc Nijmegen)	Loss of function of SETD1A causes a distinct neurodevelopmental disorder in humans, whereas knock down of the orthologue Set1 shows impaired memory in Drosophila
14.15 - 14.30	Wout Weuring (UMC Utrecht)	Efficacy of voltage-gated sodium channel subtype selective compounds in a novel zebrafish model for Dravet syndrome
14.30 - 14.45	Juliette Kamp (LUMC Leiden)	BRCA1-associated mutational signatures are a consequence of polymerase Theta-Mediated End-Joining
14.45 - 14.55	Discussion and closure	

14.55 - 15.30	**Coffee break - Pitch & Play by Eppendorf**	**Foyer & Room 4**

EX VIVO AND PATIENT-SPECIFIC MODELS — Room 2

15.30 - 15.45	Michaela Bartusel (University of Cologne)	Modelling the pathological long-range regulatory effects of structural variation in the neural crest with patient-specific hiPSC
15.45 - 16.00	Cedric Thues (KU Leuven)	Unravelling the pathophysiology of a novel tubulinopathy: Circumferential Skin Creases syndrome, Kunze type
16.00 - 16.15	Elke de Boer (Radboudumc Nijmegen)	Aberrant accumulation and mislocalization of ANKRD11 in the nucleolus of patient-derived cells indicate a dominant-negative mechanism underlying the phenotype in KBG syndrome
16.15 - 16.30	**Power break**	
16.30 - 16.45	Ronja Hollstein (University of Bonn)	Establishing human neural crest cells to functionally characterize risk loci associated with common facial disease
16.45 - 17.00	Frederic Thieme (University of Bonn)	Analyzing non-coding risk variants for non-syndromic cleft lip with/without cleft palate using massively parallel reporter assays (MPRAs)
17.00 - 17.15	Lise van Wijk (LUMC Leiden)	Functional analysis of BRCAness in female cancers: translation to clinical applications
17.15 - 17.30	Jeroen Meekels (Maastricht UMC+)	Development of an innovative method for comprehensive preimplantation genetic testing
17.30 - 17.40	Discussion and closure	

18.00 - 20.00	**Walking dinner**	**Foyer & Room 4**
20.00 - 21.00	*Prof. Thierry Voet (KU Leuven)*	Single-cell multi-omics to study DNA mutation, genetic heterogeneity and disease
21.00 - 23.00	**Social Evening**	**Boerenkelder**

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07.00 - 09.00 **Breakfast** — **Grote Eetzaal**

COMPLEX GENETICS — Room 2

09.00 - 09.15	Julia Schröder (University Hospital Bonn)	Functional follow-up of 5p15 risk locus associated with Barrett's esophagus and esophageal adenocarcinoma
09.15 - 09.30	Mandy Meijer (Radboudumc Nijmegen)	The interaction between genetics and the environment in ADHD persistence and impulsive and aggressive behavior
09.30 - 09.45	Julia Welzenbach (University of Bonn	Integration of genetic and functional data reveals insights into biological effects in craniofacial development and orofacial clefting
09.45 - 10.00	Roel van Reij (Maastricht University)	Genome-wide association analysis identifies potential risk locus for chronic postsurgical pain
10.00 - 10.15	Philip Jansen (VUmc Amsterdam)	Population genetics in the Big Data era: results from large scale GWAS of behavioral traits
10.15 - 10.25	Discussion and closure	

10.25 - 11.00 **Coffee break - Pitch & Play by Sanofi Genzyme** — **Foyer & Room 4**

GENOME STABILITY, STRUCTURAL VARIATION AND DIAGNOSTICS — Room 2

11.00 - 11.15	Luca Magdalena Schierbaum (University Hospital Bonn)	Copy number variation analysis in patients with anorectal malformation and cleft lip and palate in order to determine 22q11.2 mircodeletion and -duplication frequencies and identify rare disease-causing CNVs
11.15 - 11.30	Lisanne Vervoort (KU Leuven)	Optical Mapping of 22q11.2 Low Copy Repeats reveals structural hypervariability
11.30 - 11.45	Vyne van der Schoot (Maastricht UMC+)	1 in 38 individuals at risk of a dominant medically actionable disease
11.45 - 12.00	Margot van Riel (KU Leuven)	Non-invasive prenatal diagnosis using cervical trophoblast cells
12.00 - 12.15	Jenny Singh (LUMC Leiden)	A novel Zinc Finger protein in DNA double-strand break repair
12.15 - 12.25	Discussion and closure	

12.25 - 13.40 **Lunch & Award ceremony by Alexander Hoischen** — **Grote Eetzaal**

13.40 - 13.50 **Photo shoot prize winners & all participants** — **Enclosed garden**