

PROGRAMME GENETICS RETREAT

JOINT MEETING BESHG/NVHG



WEDNESDAY 20 APRIL 2022

10.30 - 11.00	Registration & welcome coffee		Catering area
11.00 - 11.15	Opening		Plenary A
#1 LESSON'S FROM RARE DISEASES: FROM GENE IDENTIFICATION TO FUNCTIONAL UNDERSTANDING AND THERAPIES			Plenary A
11.15 - 11.30	A structural variant of the C-terminal prion-like domain of TDP-43 causes vacuolar muscle degeneration	Pedro Ervilha Pereira (Gent)	
11.30 - 11.45	CLEC16A mislocalization and impaired interaction with the retromer underly a recessive severe neurodevelopmental disorder.	Daphne Smits (Rotterdam)	
11.45 - 12.00	ATRIP-deficient patient expands molecular and clinical spectrum of Seckel syndrome	Lynn Backers (Gent)	
12.00 - 12.15	Novel mRNA therapy restores GALT protein and enzyme activity in young classic galactosemia zebrafish	Britt Delnoy (Maastricht)	
12.15 - 12.30	Discussion and closure session #1		
12.30 - 13.30	Lunch		Catering area
#2 APPLICATION OF LATEST TECHNOLOGIES IN HUMAN GENETICS			Plenary A
13.30 - 13.45	FiberFISH mapping of 22q11.2 rearrangements shows locus heterogeneity	Ruben Cools (Leuven)	
13.45 - 14.00	Objective 3D facial phenotyping in Cri-du-Chat Syndrome	Michiel Vanneste (Leuven)	
14.00 - 14.15	Deciphering the genetic cause of recurrent and spontaneous pregnancy loss	Rick Essers (Maastricht)	
14.15 - 14.30	Identification of potential core functions in human immune responses of genes with previously unknown function	Emil Vorsteveld (Nijmegen)	
14.30 - 14.45	Variant effect prediction based on custom long-read transcriptomes	Renée Salz (Nijmegen)	
14.45 - 15.00	Discussion and closure session #2		
15.30 - 16.00	Coffee break		Catering area
#3 APPLICATION OF LATEST TECHNOLOGIES IN HUMAN GENETICS			Plenary A
15.30 - 15.45	Diagnostic yield of partial exome sequencing in 872 children with neurodevelopmental disorders	Sebastian Neuens (Brussels)	
15.45 - 16.00	The performance of GS as a first-tier test for neurodevelopmental disorders	Bart van der Sanden (Nijmegen)	
16.00 - 16.15	Mutational processes in childhood acute lymphoblastic leukemia	Cédric van der Ham (Utrecht)	
16.15 - 16.45	Energizer		Terrace
#4 COMMON VARIANT EFFECTS			Plenary A
16.45 - 17.00	Impact of common genetic variants on cytokine response heterogeneity upon BCG vaccination in infants from Guinea-Bissau	Collins Boahen (Nijmegen)	
17.00 - 17.15	Genome-wide and pathway-specific polygenic risk scores in an Alzheimer's disease case-control cohort	Yasmina Abakkouy (Leuven)	
17.15 - 17.30	Discussion and closure sessions #3 and #4		
17.30 - 18.00	Aperitif		Catering area
18.00 - 19.30	Dinner		Catering area
19.30 - 20.30	Keynote lecture by Musa Mhlanga		Plenary A
20.30 - 21.00	Discussion and wrapping up		
21.00 - 23.00	Social Evening		Brouwerij De Halve Maan