JOINT MEETING

UK / Dutch Clinical Genetics Societies & Cancer Genetics Groups

Joint Meeting 2018 | 12 & 13 March - Utrecht, the Netherlands

Program *(Changes reserved) 20180131*

General theme: "Future proof clinical genetics"

Day 1: Monday 12 March – ***Joint program***

09:00	Registration and coffee
09:30	**Opening address**
	Dr Frederik Hes, chairman of the Dutch society of Clinical Genetics
09:40	**Kick off presentation**
	Professor Annelien Bredenoord, Utrecht
10:00	**title will follow, presentation on genetics, ethics and law**
	Professor Anneke Lucassen, Southampton
10:20	**Rise of the Robots and Superomics for All**
	Professor Rolf Sijmons, Groningen
10:40 – 11:10	**BREAK**
11:10 – 12:25	**SYMPOSIUM: Future proof 'testing and interpretation'**
	Chairs: *Dr Frederik Hes, Leiden & Dr Jane Hurst, London*
	SUBMITTED ABSTRACTS
11:10	Rapid whole exome sequencing in clinical care – when to and when not to use this diagnostic tool
	Chantal Deden, Nijmegen
11:25	Rapid whole exome sequencing; implementation in the prenatal setting
	Ilse Feenstra, Nijmegen
11:40	Making the most of prenatal exome sequencing: start of a national database
	Gijs Santen, Leiden
11:55	Copy Number Variants (CNVs) Affecting Cancer Predisposing Genes (CPGs) Detected As Incidental Findings In Routine Germline Diagnostic Chromosomal Micro-Array (CMA) Testing
	Emma Woodward, Manchester
12:10	Segregation analysis in genomic variant interpretation
	Jan-Maarten Cobben, Amsterdam
12:25	Genome-wide meta-analysis of insomnia (N=1,300,000) identifies novel genes and functional pathways
	Philip Jansen, Amsterdam
12:40 – 14:00	**LUNCH and POSTER VIEWING**
13:40 – 14:00	**CGS General meeting**
14:00 – 15:30	**SYMPOSIUM: Theme Mendelian disorders and beyond**

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	Chairs: *Dr Tjitske Kleefstra, Nijmegen & Dr Francesca Faravelli, London*
	SUBMITTED ABSTRACTS
14:00	Extracting pharmacogenetic genotypes from exome data; benefits and challenges *Marjolein Kriek, Leiden*
14:15	The Role of Detailed Clinical Phenotyping in Genomic Medicine *Hannah Titheradge, Birmingham*
14:30	Molecular dissection of complex germline structural variation using a multi-omics approach *Sjors Middelkamp, Utrecht*
14:45	Successful use of social media in the study of rare disorders: The Chromosome 6 Project *Aafke Engwerda, Groningen*
15:00	Preconception carrier screening for multiple disorders: evaluation of a screening offer in a Dutch founder population *Inge Mathijssen, Amsterdam*
15:15	A couple-based approach to expanded preconception carrier screening *Juliette Schuurmans, Southampton & Groningen*
15:30 – 16:00	**BREAK**
16:00 – 17:40	**SYMPOSIUM: Syndromal Haematogenetics**
	Chairs: *Dr Mieke van Haelst, Amsterdam & Dr Helen Hanson, London*
	INVITED SPEAKERS:
16:00	Hereditary germline mutations and developmental disorders in the haematological malignancy genes *Dr Kate Tatton Brown, London*
16:20	Myelodysplasia and acute myeloid leukaemia: syndromes and susceptibilities *Professor Sahar Mansour, London*
16:40	**A multitude of mosaics – Carter lecture** *Professor Andrew Wilkie, Oxford*
17:40	**DRINKS**
18:30 – 22:00	**CONFERENCE DINNER + PARTY**

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Day 2: Tuesday 13 March – *Parallel Program Clinical genetics*

08:30	Registration and coffee
09:00 – 10:50	**SYMPOSIUM: Theme neurogenetics**
	Chairs: *Dr Grazia Mancini, Rotterdam & TBA*
09:00	**INVITED SPEAKER:**
	TBA
	SUBMITTED ABSTRACTS
09:20	Mosaicism of de novo pathogenic SCN1A variants in epilepsy is a frequent phenomenon that correlates with variable phenotypes
	Iris de Lange, Utrecht
09:35	RANBP2 mutations lead to acute necrotizing encephalopathy type 1 – the Dutch experience
	Marrit Hitzert, Groningen
09:50	Genetic and phenotypic findings in five individuals with de novo mutations in ZBTB18
	Rhian Thomas, London
10:05	Genetic Epidemiology of Familial Motor Neuron Disease in the Netherlands
	Charlotte de Bie, Utrecht
10:20	Exploring Genetic Predisposition to Foetal Anticonvulsant Syndromes (FACS)
	Adam Jackson, Manchester
10:35	Insights into MED13L syndrome: A report of 53 patients with SNVs in MED13L and proposal for diagnostic criteria
	Rhoda Akilapa, London
10:50 – 11:15	**BREAK**
11:15 – 12:35	**SYMPOSIUM: Theme phenotypes**
	Chairs: *Dr Paul van der Zwaag, Groningen & TBA*
	INVITED SPEAKER:
11:15	The DDD study - Demonstrating the value of iterative analysis of NGS data
	Dr Helen Firth, Cambridge
	SUBMITTED ABSTRACTS
11:35	The zebrafish as a model for cardiac abnormalities in Cantú Syndrome
	Helen Roessler, Utrecht
11:50	Recessive ASNA1 mutations cause severe pediatric dilated cardiomyopathy and early death
	Myrthe van den Born, Rotterdam
12:05	Determination of underlying genetic variations in an obese cohort of more than 1000 patients and the effect of bariatric surgery
	Mellody Cooiman, Arnhem

12:20	Genetic studies of cases with Bohring Opitz and Bohring Opitz like syndrome associated with ASXl1 and KLHL7 mutations *Ruth Newbury-Ecob, Bristol*
12:35 – 13:45	**LUNCH and POSTER VIEWING**
13:45 – 15:15	**SYMPOSIUM: Theme phenotypes (continued)** Chairs: *TBA* **SUBMITTED ABSTRACTS**
13:45	Novel syndrome due to de novo missense mutations of CHD3 in 35 patients *Lot Snijders Blok, Nijmegen*
14:00	Changes in the urinary extracellular vesicle proteome are associated with nephronophthisis-related ciliopathies *Marijn Stokman, Utrecht*
14:15	KAT6A Syndrome: International Collaboration and Patient/Family Participation Expand our Knowledge of Phenotype and Genotype *Joanna Kennedy, Bristol*
14:30	Four further patients with CTNND1 mutations: new insights into the phenotype of Blepharocheilodontic syndrome? *Athina Ververi, London*
14:45	ARID1B in Coffin-Siris Syndrome and Non-Syndromic Intellectual Disability *Eline van der Sluijs, Leiden*
15:00	An inactivating mutation in the histone deacetylase SIRT6 causes multiple congenital anomalies and perinatal mortality *Merel van Maarle, Amsterdam*
15:15 – 15:30	Time to switch rooms
15:30 – 16.30	**JOINT CLOSURE & AWARD CEREMONY** **Ben ter Haar Prize & Lecture** **Award ceremonies:** **Dutch and UK presentations SpR & Robin Winter Prize** Results of the jury **Poster prize** Results of the public vote **Conference closure** *Dr Frederik Hes,* chairman of the Dutch society of Clinical Genetics
16:30	End of the conference

Day 2: Tuesday 13 March – *Parallel Program Cancer genetics*

08:30	Registration and coffee
09:00 – 10:45	**SYMPOSIUM: Theme Screening and prevention**
	Chairs: *Dr Margreet Ausems, Utrecht & TBA*
	SUBMITTED ABSTRACTS
09:00	PMS2-associated Lynch syndrome: the odd one out
	Sanne ten Broeke, Leiden
09:15	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): clinical and molecular features, and renal cancer surveillance imaging
	Claire Forde, Manchester
09:30	Oral contraceptive use and breast cancer risk: retrospective and prospective analyses from a BRCA1 and BRCA2 mutation carrier cohort study
	Lieske Schrijver, Amsterdam
09:45	Clinical aspects of SDHA-related pheochromocytoma and paraganglioma; a nationwide study
	Frederik Hes, Leiden
10:00	Renal imaging in Dutch patients with Birt-Hogg-Dubé syndrome: Screening compliance and outcomes
	Irma van de Beek, Amsterdam
10:15	The LiFe-Guard study: Impact of surveillance with whole body MRI in a Dutch Li-Fraumeni Syndrome cohort
	Marielle Ruijs, Amsterdam
10:30	Genetic counselling and testing of over 350 young women with breast cancer for Li-Fraumeni Syndrome in the Netherlands: nationwide overview of laboratory practice and survey of genetics professionals' experiences and attitudes
	Jette Bakhuizen, Utrecht
10:45	**BREAK**
11:15 – 12:35	**SYMPOSIUM: Theme New developments and new pathways**
	Chairs: *TBA*
	INVITED SPEAKERS:
11:15	Somatic sequencing in the investigation of hereditary cancer predisposition
	Dr Andrew Wallace, Manchester
11:35	Hereditary cancer predisposition and the interplay between tumor and germline testing
	Professor Marjolijn Ligtenberg, Nijmegen
	SUBMITTED ABSTRACTS
11:55	A comparison of genetic testing criteria: sensitivity, clinical benefits and VUS rate

	Megan Berkeley, Dublin
12:10	Transformative local pathways for cancer genomics
	Katie Snape, London
12:25	Germline sequencing of advanced prostate cancer patients in the BARCODE2 trial
	Sarah Benafif, London
12:40 – 13:45	**LUNCH and POSTER VIEWING**
13:45 – 15:15	**SYMPOSIUM: Theme Further defining the phenotype of known cancer predisposition genes**
	Chairs: *TBA*
	SUBMITTED ABSTRACTS
13:45	Inherited BRCA1 epimutation as a novel cause of breast and ovarian cancer
	Gareth D Evans, Manchester
14:00	A novel mainstreaming model for Lynch syndrome genetic testing in colorectal cancer patients
	Bianca Desouza, London
14:15	Future proofing genetic counselling for cancer genetics in the genomics era
	Kelly Kohut, London
14:30	Individuals with biallelic germline NTHL1 mutations are at high risk for a wide variety of cancers
	Nicoline Hoogerbrugge, Nijmegen
14:45	CDC73-related disorders: clinical manifestations and case detection in primary hyperparathyroidism
	Karin van der Tuin, Leiden
15:00	Germline pathogenic variants in PALB2 and other cancer-predisposing genes in CDH1-negative diffuse gastric cancer families
	Ellie Fewings, Cambridge
15:15 – 15:30	Time to switch rooms
15:30 – 16.30	**JOINT CLOSURE & AWARD CEREMONY**
	Ben ter Haar Prize & Lecture
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	Results of the jury
	Poster prize
	Results of the public vote
	Conference closure
	Dr Frederik Hes, chairman of the Dutch society of Clinical Genetics
16:30	End of the conference