

Joint Dutch/UK Clinical Genetics Societies and Cancer Genetics Groups meeting 2020

Day 1 - Monday February 10th

08:45 Registration

09:30 Opening address - *Frances Elmslie*, chair of the UK CGS

09:35 Psychological impact of WGS - ***Saskia Sanderson***

10.05-10.35 Dutch geneticists in the UK- what are the main differences and what can we learn from each other? - ***Marijke Wevers***

UK experience of the Netherlands - ***Anneke Lucassen***

10:35 – 11:00 BREAK

11.00 Martijn Breuning lecture

Polygenic risk scores - ***Cecile Janssens***

11.50 – 12.50 SUBMITTED ABSTRACTS - Session 1

11.50 Genome sequencing provides insights into tumour diversity in CYLD cutaneous syndrome
Helen R. Davies, University of Cambridge, UK

12:05 Strategies to increase diagnostic yield from the 100,000 genomes project. *Meriel McEntagart, St George's University Hospitals NHS Foundation Trust*

12:20 Beyond no primary findings: what we miss, why, and how to find it. *Eleanor Hay, Great Ormond Street Hospital, London, UK*

12:35 Evaluating the contribution of near-splice variants to rare disease in the 100,000 Genomes Project. *Alexander Blakes, University Hospitals Southampton NHS Foundation Trust*

12.50 – 14:00 LUNCH and POSTER VIEWING - Session A (even numbers)

12.50 – 13:10 CGS General meeting

14:00 – 15:30 SYMPOSIUM 1 Treatments for genetic conditions

14:00- 14:30 Treatments in Tuberous Sclerosis - ***Nick Annear***

14:30 – 16:00 SUBMITTED ABSTRACTS - Session 2

14:30 Clinical improvement after treatment in a girl with compound heterozygous mutations in SLC5A6 *Saskia N. van der Crabben, Clinical Genetics, Amsterdam, NL*

14.45 Development & Implementation of a Point-of-care Pharmacogenetic Test to Avoid Antibiotic Related Hearing Loss in Neonates. *John H. McDermott, Manchester Centre for Genomic Medicine, UK*

15:00 The intervention in PHOspolamban Related Cardiomyopathy Study (i-PHORECAST), *Wouter P. te Rijdt, University of Groningen, NL*

15:15 The role of in vivo metabolomics in hereditary cancer syndromes caused by citric acid cycle gene mutations *Ruth T. Casey, University of Cambridge, UK*

15:30 The coding microsatellite mutation profile of PMS2-deficient colorectal cancer; one step closer towards unraveling the mystery behind gene-specific tumorigenesis in Lynch syndrome *Sanne W. ten Broeke, University Medical Center Groningen, NL*

15:45 Progress with PREGCARE: personalised evaluation of recurrence risk after birth of a child with a de novo mutation *Andrew Wilkie, University of Oxford, UK*

16:00 - 16:30 BREAK

16:30 – 17:30 The Carter lecture - *Helen Firth*

17:30 - 18:30 Reading from *The Phlebotomist* By *Ella Road*

18:30 DRINKS

19:30 – 23.30 CONFERENCE DINNER + PARTY

Day 2: Tuesday February 11 – Parallel Program Clinical genetics

08:30 REGISTRATION

09:00 – 11:15 SYMPOSIUM 2 Management of neurogenetic disorders

9:00 Genetics of Hypomyelination: New Insights - *Nicole Wolf*

9:30 Treatment strategies in the human type I interferonopathies - *Yanick Crow*

10:00 Management of epileptic encephalopathies - *Elaine Hughes*

10.30 – 11.15 SUBMITTED ABSTRACTS Session 3

10:30 Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform specific start-loss mutations of essential genes can cause genetic diseases. Tahsin Barakat, *Erasmus MC University Medical Center, Rotterdam, NL*

10.45 – 11:15 BREAK

11:15 – 12:45 SUBMITTED ABSTRACTS Session 4

11:15 Understanding the Phenotypic Spectrum of Synaptic Vesicle Cycling Disorders. *Kate Baker, University of Cambridge*

11:30 Exonic *ERBB4* deletions cause non-syndromic intellectual disability or epilepsy
Zerin Hyder, Manchester Centre for Genomic Medicine, Manchester, UK

11:45 Variants in peptidyl prolyl isomerase-like protein 1 (PPIL1) cause a novel brain malformation syndrome. *Eamonn Sheridan, Leeds Institute for Molecular Medicine, UK*

12:00 Biallelic variants in *COPB1* cause a novel severe intellectual disability syndrome with variable presence of microcephaly and cataracts. *William Macken, Wessex Clinical Genetics Service, Southampton, UK*

12:15 GWAS of brain volume on 47,316 individuals and cross-trait analysis with intelligence identifies shared genomic loci and genes. *Philip R Jansen, Vrije Universiteit Amsterdam, NL*

12:30 Gene-environment interaction study suggests significant interaction between gestational age at birth and genetic risk for psychiatric disease on cognition at age four. *Harriet Cullen, Centre for the Developing Brain, Kings College, London*

12:45 – 13:45 LUNCH and POSTER VIEWING - Session B (odd numbers)

13:45 – 15:15 SYMPOSIUM 3 _ Prenatal and Postnatal Dysmorphology

SUBMITTED ABSTRACTS Session 5

13:45 Characterisation of clinical and molecular genetic spectrum of SOX11 syndrome. *Alisdair McNeill, University of Sheffield.*

14:00 Distinctive codon-specific phenotypes of *SETD2*; an international reverse-phenotyping collaboration. *Lara Menzies, Great Ormond Street Hospital, UK*

14:15 Three unrelated Dutch patients with the same homozygous OSGEP gene mutations and a relatively mild Galloway-Mowat syndrome phenotype. *Dennis Bos, University Medical Center, Groningen, NL.*

14:30 Comprehensive data from an international collaborative study on ASXL3-related disorder: 70 patients and the number keeps growing! *Schaida Schirwani, University of Sheffield*

14:45 Rapid exome sequencing in fetuses showing ultrasound abnormalities: challenges in the implementation in prenatal diagnosis. *Nicole Corsten-Janssen, University of Groningen, NL*
15:00 Prenatal Diagnosis in the Era of Genomic Medicine. *Suzanne Drury, Congenica Ltd*

15:15 – 15:45 Tea

15:45 – 16.30 JOINT CLOSURE & AWARD CEREMONY

Ben ter Haar Prize & Lecture

Dutch and UK presentations SpR & Robin Winter Prize
Results of the jury

Poster prize
Results of the public vote

Conference closure *Frances Elmslie*

16:30 End of the conference

We wish you all a safe journey home and hope to meet again in 2022 for another inspiring conference

Day 2: Tuesday February 11 – *Parallel Program Clinical genetics*

08:30 REGISTRATION

09:00 – 10:45 SUBMITTED ABSTRACTS session 6

- 09:00 High detection rate from cancer gene panel testing/exome sequencing in BRCA-negative women with familial ovarian cancer *Nicola Flaum, University of Manchester, UK*
- 09:15 Experiences with transition of gene panel DNA diagnostics from clinical geneticists to treating physicians in breast cancer patients Maaïke L. Haadsma, *Radboud University Medical Center, Nijmegen, NL*
- 09:30 The contribution of germline pathogenic variants beyond BRCA1/2/PALB2 to contralateral breast cancer in women with a younger onset breast cancer, *Alexey Larionov, University of Cambridge, UK*
- 09:45 High likelihood of actionable pathogenic variants in breast cancer genes in women with very early onset breast cancer; but little gain from additional panel testing, *D. Gareth Evans, University of Manchester, UK*
- 10:00 Clinical findings in Dutch Families with BAP1-Tumor Predisposition Syndrome C. Chau, *Leiden University Medical Center, NL*
- 10:15 Renal cell carcinoma (RCC) predisposition genes: Results from a clinical service evaluation and the 100 000 Genomes Project. *A. Andreou, University of Cambridge, UK*
- 10:30 European Reference Network on rare genetic tumour risk syndromes: What is in it for me? *Nicoline Hoogerbrugge, Radboud University Medical Center, Nijmegen, NL*

10:45 – 11:15 BREAK

11:15 – 12:40 SYMPOSIUM 4: Personalized medicine in tumor syndromes

INVITED SPEAKERS:

- 11:15 Personalised medicine in BRCA1/2 carriers – *Jolien Tol*
- 11:35 Personalised medicine in bowel cancer - *Nirupa Murugaesu*

11.55 – 12.40 SUBMITTED ABSTRACTS session 7

- 11:55 Cancer prevention with aspirin and with resistant starch in people with Lynch syndrome *John Burn, University of Newcastle, UK*
- 12:10 Clinical and molecular assessment for Familial Adenomatous Polyposis in a cohort of patients with Desmoid Fibromatosis *Elena Cojocaru, Royal Marsden NHS Foundation Trust, UK*
- 12:25 The complexities of analysis for APC mosaicism, *Maartje Nielsen, Leiden University Medical Centre, NL*

12:40 – 13:45 LUNCH and POSTER VIEWING Session B (odd numbers)

13:45 – 15:15 SUBMITTED ABSTRACTS session 8

- 13:45 **UKCGG Essay Prize** Should All Individuals Be Screened for Genetic Predisposition to Cancer? *Sarah Wedderburn, Glasgow, UK*
- 14:00 Clinical characteristics and outcome of children with WAGR syndrome and Wilms tumor treated according to SIOP-RTSG protocols *Janna A. Hol, Princess Máxima Center for Pediatric Oncology, Utrecht, NL*

- 14:15 EHTG Delineating the cancer risks of MSH6 mutation carriers – preliminary results with simple statistics, *A.S. van der Werf – 't Lam, Leiden University Medical Center, NL*
- 14:30 Impact of aneuploidy and chromosomal instability in shaping the renal cell carcinoma immune landscape *Laia Bassaganyas, University of Cambridge, UK*
- 14:45 Acute myeloid leukaemia associated with ataxia telangiectasia: complex karyotype with wildtype TP53 and mutant KRAS, G3BP1 and IL7R *Mae A. Goldgraben, University of Cambridge, UK*
- 15:00 Screening Strategies for Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC): A Cost-Effectiveness Analysis *Alexander J. Thompson, University of Manchester, Manchester, UK*

15:15 – 15:45 Tea

15:45 – 16.30 JOINT CLOSURE & AWARD CEREMONY

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Poster prize

Results of the public vote

Conference closure

Dr Frances Elmslie

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Dysmorphology Club Wednesday Feb 12th