

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

Day 1 - Monday February 10th

VENUE: Francis Crick Auditorium

08:45 Registration

09:30 Welcome – *Dr Frances Elmslie, President, UK Clinical Genetics Society*

JOINT SESSION 1

Chairs: *Dr Frances Elmslie, London, UK and Dr Wendy van Zelst-Stams, Amsterdam, NL*

09:35 Psychological and behavioural reactions to receiving personal genomic information –
Dr Saskia Sanderson, University College London, UK

10:05 Dutch geneticists in the UK- what are the main differences and what can we
learn from each other? – *Dr Marijke Wevers, Nijmegen, UK*

10:20 Double Dutch or plain English – Reflections on NL/UK genetics practices from an
anglicised Dutch person – *Professor Anneke Lucassen, University of Southampton,
UK*

10:35 – 11:00 BREAK

11:00 **Martijn Breuning lecture**
Polygenic risk scores: going where? – *Professor A. Cecile J.W. Janssens, Emory
University, USA*

11:50 – 12:50 **SUBMITTED ABSTRACTS - Session 1**

Chairs: *Dr Marc Tischkowitz, Cambridge, UK and Professor Han Brunner, Nijmegen, NL*

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**

Chairs: *Dr Andrew Douglas, Southampton, UK and Dr Maartje Nielsen, Leiden, NL*

14:00- 14:30 Treatments in Tuberous Sclerosis Complex (TSC): From bench to bedside.. to
complex multi-disciplinary service - *Dr Nicholas Annear, St George's University Hospitals NHS
Foundation Trust, London, UK*

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 PHOspholamban Related Cardiomyopathy Study (i-PHORECAST), *Wouter P. te Rijdt, University of Groningen, NL*
- 15:15 High detection rate from cancer gene panel testing/exome sequencing in BRCA-negative women with familial ovarian cancer. *Nicola Flaum, University of Manchester, 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 – Data-sharing for Discovery and Diagnosis - *Dr Helen Firth, Cambridge University Hospitals NHS Foundation Trust, UK*
Introduction - *Dr Frances Elmslie*

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

18:30 WINE RECEPTION
Conference Centre

19:30 – 21:30 CONFERENCE DINNER
Hinxton Hall

21:30 – 23:30 PARTY with music from the Altitudes
Conference Centre

Day 2 - Tuesday February 11

Parallel Programme - Clinical Genetics

VENUE: Francis Crick Auditorium

08:30 REGISTRATION

09:00 – 10:45 SYMPOSIUM 2 - Management of neurogenetic disorders

Chairs: Professor Eamonn Sheridan, Leeds, UK and Dr Arjan Bouman, Amsterdam, UK

9:00 Hypomyelinating leucodystrophies – lessons from genetics – **Dr Nicole Wolf, VU University Medical Center, Amsterdam, UK**

9:30 Treatment strategies in the human type I interferonopathies – **Professor Yanick Crow, University of Edinburgh, UK**

10:00 Treating complex epilepsies in children: managing clinician and families' expectations – **Dr Elaine Hughes, King's College Hospital and Evelina London Childrens' Hospital, UK**

10:30 – 10:45 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. Stefan Barakat, *Erasmus MC University Medical Center, Rotterdam, NL*

10:45 – 11:15 BREAK

11:15 – 12:45 SUBMITTED ABSTRACTS Session 4

Chairs: Dr Jane Hurst, London, UK and Dr Nicole Wolf, Amsterdam, NL

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 SUBMITTED ABSTRACTS Session 5

Chairs: Meena Balasubramaniam, Sheffield, UK and Professor Diana Baralle, Southampton, UK

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

VENUE: Francis Crick Auditorium

15:45 – 16:30 JOINT CLOSURE & AWARD CEREMONY

Chairs: Dr Frances Elmslie & Dr Bernadette van Nestelrooy

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 Dr 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 11th February
Parallel Programme - Cancer Genetics

VENUE: Rosalind Franklin Pavilion

08:30 REGISTRATION

09:00 – 10:45 SUBMITTED ABSTRACTS Session 6

Chairs: Dr Helen Hanson, London, UK and Christi van Asperen, Leiden, NL

- 09:00 The role of in vivo metabolomics in hereditary cancer syndromes caused by citric acid cycle gene mutations. *Ruth T. Casey, University of Cambridge, UK*
- 09:15 Experiences with transition of gene panel DNA diagnostics from clinical geneticists to treating physicians in breast cancer patients. *Maike 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. *Elke van Veen, 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 3: Personalised medicine in tumour syndromes

Chairs: Dr Ian Frayling, Cardiff, UK and Dr Marleen Kets, Nijmegen, NL

- 11:15 BRCA 1/2 carriers with breast cancer: tailored treatment? – **Dr Jolien Tol, Jeroen Bosch Hospital, NL**
- 11:35 The 100,000 Genomes Project: Update on the Cancer Programme, whole genome sequencing & the impact on Oncology – **Dr Nirupa Murugaesu, St George's University Hospitals, London, UK**

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

Chairs: Professor Julian Barwell, Leicester, UK and Prof Nicoline Hoogerbrugge, Nijmegen, NL

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. *Emma Woodward, University of Manchester, UK*

15:15 – 15:45 Tea

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