**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 – ***A. 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*

12:35Evaluating 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 Complex - ***Nicholas Annear***

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

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

14.45Development & 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*

15:15The 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 Programme - 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 – 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. 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** **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 11th February**

***Parallel Programme - Cancer 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. *Maaike 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 3: Personalised medicine in tumour 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, UK*

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