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
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 Mathijsen, 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
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 ASX1 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
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12:10  Megan Berkeley, Dublin
Transformative local pathways for cancer genomics

12:25  Katie Snape, London
Germline sequencing of advanced prostate cancer patients in the BARCODE2 trial

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

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16:30  End of the conference