JOINT MEETING

UK / Dutch Clinical Genetics Societies & Cancer Genetics Groups

# The 4th Joint Spring Conference

**7-9 March 2016**

**The City Hall, Cardiff Civic Centre**

## Day 1: Monday 7th March – Joint Programme

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| **09:00** | **REGISTRATION & COFFEE** | **Lower Hall** |
| **09:50** | **Welcome/ Introduction**- Professor Dhavendra Kumar | **Assembly Room** |
| **10:00** | **Opening address/ Key note Lecture**<br>Professor Ruth Newbury-Ecob, President Clinical Genetics Society, UK | |
| **10:30 – 13:00** | **JOINT SESSION I** **Lead- Clinical Genetics**<br>**Rare Diseases – Recent Trends in Diagnosis and Therapy**<br>***Chairs: Professor Angus Clarke,*** *Cardiff, UK* & ***Dr Mieke van Haelst,*** *Uttrecht, NL* | **Assembly Room** |
| **10:30** | **Diagnostic potentials of Whole Exome & Whole Genome Sequencing (WES/WGS) for Rare Genetic Diseases**<br>Professor Han Brunner, Nijmegen & Maastricht, NL | |
| **11:00** | **Visualising phenotypic and genomic relationships with DECIPHER – A community endeavour to map the Clinical Genome** Helen Firth, Cambridge, UK | |
| **11:15 – 11:45** | **Coffee Break & Poster Viewing** | **Lower Hall** |
| **11:45** | **Classification and clinical management of Variants of Uncertain Significance in high penetrance cancer predisposition genes** Setareh Moghadasi, Leiden, NL | |
| **12:00** | **New drug developments for rare genetic diseases- the model of type 1 Interferonopathies**<br>Professor Yanick Crow, Manchester, UK/Marseille, France | |
| **12:30** | **From Gene Association to Medicine**<br>Dr Jonathan Appleby, London, UK | |
| **13:00** | **Annual General Meeting: CGS - UK** | |
| **13:00-14:00** | **LUNCH & POSTER VIEWING** | **Lower Hall** |
| **14:00 – 15:45** | **JOINT SESSION II** **Lead - Cancer Genetics**<br>**Neoplasia and Malformations**<br>***Chairs: Dr Lucy Side,*** *London, UK* ***& Dr Margreet Ausems,*** *Utrecht, NL*<br>**14:00 Update on cancer predisposition in childhood**<br>Dr Marjolijn Longmans, Leiden, NL<br>**14.25 Molecular basis of neoplasia in malformations- the paradigm of Wilms Tumour**<br>Dr Keith Brown, Bristol, UK<br>**14:50 Dysmorphic syndromes with Wilms Tumor-a clinical and molecular overview**<br>Dr Marry van den Heuvel-Eibrink, Utrecht, NL<br>**15:15 Selectively targeting TSC1/2 deficient cells by exploiting endoplasmic reticulum stress,** Mark Davies, Swansea/Cardiff, Wales, UK<br>**15:30 High yield of causative mutations by whole exome sequencing in selected individuals with childhood cancer**, Ilja Diets, Nijmegan, NL | **Assembly Room** |
| **15:45 – 16:15** | **TEA BREAK & POSTER VIEWING** | **Lower Hall** |

**16:15 – 18:30** **JOINT SESSION III:**

**Clinical & Cancer Genetics Trainee Presentations**

***Chairs: Dr Jane Hurst, London, UK & Professor Nine Knoers, Utrecht, NL***

**With Judges Panel**- Joint UK Dutch Clinical (2) & Cancer Groups (2)

**UK- THE ROBIN WINTER PRIZE + UK/ DUTCH PRIZES (5 prizes)**

**16:15** **Experience of participation in a therapeutic drug trial for neonatal patients with X-linked Hypohidrotic Ectodermal Dysplasia (XLHED),** Arveen Kamath, Cardiff, UK

**16:30** **Heterozygous KIDINS220/ARMS nonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity,** Glen Monroe, Utrecht, NL

**16:45** **Compound heterozygous NEK1 variants in two siblings with oral-facial-digital syndrome type II (Mohr syndrome),** Marijn F. Stokman, Utrecht, NL

**17:00** **Imprinting: the Achilles heel of trio-based Exome sequencing ,** Gijs Santen, Leiden, NL

**17:15** **The Development of a Clinical Screening Instrument for Tumor Predisposition Syndromes in Childhood Cancer Patients: protocol for a prospective, observational, multi-center study (TuPS)**, Saskia Hopman, Amsterdam, NL

**17:30** **SMC1A truncating mutations are associated with a severe epilepsy phenotype which is distinct from de Lange syndrome**, Suresh Somarathi, Manchester, UK

**17:45** **Risk factors for the presence of pathogenic APC and biallelic MUTYH mutations in patients with multiple adenoma**s, Sanne ten Broeke, Leiden, NL

**18:00** **Strategy to Knockout Type V Collagen Using the CRISPR-Cas9n System,** Andrea Cordaro **,** Bristol, UK

**18:15** **Characterising STAT3 signalling as a therapeutic target for vascularized tumours and Tuberous sclerosis,** Kayleigh Dodd , Cardiff, UK

**18:30** **CLOSE**

**19:45 Wine Reception and Informal Mixer/Supper at Radisson Blu Hotel, Cardiff City Centre**
**With entertainment provided by the Tenovus Choir & The Jim Barber Quartet**

## Day 2: Tuesday 8th March 2016

**08:30** **REGISTRATION & COFFEE** — Lower Hall

**09:00-11:00** **JOINT SESSION IV – PLENARY** — Assembly Room

***Chairs: Prof Ruth Newbury-Ecob,*** *Bristol, UK* ***& Dr Frederik Hes,*** *Leiden, NL*

**09:00** **Genome-wide sequencing in developmental disorders: future prospects and challenges**
Dr Matt Hurles, Cambridge, UK

**09:30** **Hartwig Whole Genome Sequencing initiative**
Dr Eric A Sistermans, Amsterdam, NL

**Keynote Address**

**10:00** **Genomics England shaping the future of clinical genetics & genomics**
Professor Mark Caulfield, London, UK

**11:00-11:30** **COFFEE BREAK** — Lower Hall

**11:30 – 13:00**

### SESSION V: CLINICAL GENETICS — Assembly Room

***Chairs: Professor Daniela Pilz,*** *Glasgow, UK* ***& Dr. Grazia Mancini,*** *Erasmus*

**11:30 Periventricular nodular heterotopias**
Dr Carlos Cardoso, INSERM, France

**12:00 Polymicrogyria- Clinical and Molecular Considerations**
Dr Andrew Fry, Cardiff, UK

**12:30 A network analysis of genes with de novo mutations in polymicrogyria patients** Katherine A Fawcett, Oxford, UK

**12:45 UNC80 mutations lead to Intellectual Disability with persistent Hypotonia, Encephalopathy, and Growth Retardation, without true Facial Dysmorphism** Jan-Maarten Cobben, Amsterdam, NL

### SESSION V: CANCER GENETICS — Ferrier Hall

***Chairs: Dr. Katie Snape,*** *London, UK* ***and Dr. Margreet Auserms,*** *Utrecht, NL*

**11:30 Contextualising the interpretation of genetic variants in common cancers**
Dr Diana Eccles, Southampton, UK

**12:00 How to deal with moderate (breast cancer) risk genes**
Dr Setareh Moghadasi, Leiden, NL

**12:30 Use of multiple SNP testing to predict breast cancer risk in a familial screening clinic** Gareth Evans, Manchester, UK

**12:45 Prostate cancer genome-wide association study from 89,000 men using the OncoArray chip identifies more than 30 novel prostate cancer susceptibility loci** Rosalind A Eeles, London, UK

**13:00 – 14:30** **LUNCH and POSTER VIEWING** — Lower Hall

**14:30 -16:00**

### SESSION VI: CLINICAL GENETICS — Assembly Room

***Chairs: Dr Angus Dobie,*** *Leeds, UK* ***& Dr Alice Brooks,*** *Erasmus, NL*

**14:30 Genetic studies in Noncompaction Cardiomyopathy using Next Generation Sequencing** L A Verlooij, Groningen, NL

**14:45 Fetal imaging in the diagnosis of skeletal dysplasias and craniosynostosis – a case series** V Ward, London, UK

**15:00 SMAD2 Mutations are associated with arterial aneurysyms and dissections** Fleur S van Dijk, Nijemegen, NL

### SESSION VI: CANCER GENETICS — Ferrier Hall

***Chairs: Professor Gareth Evans,*** *Manchester, UK* ***& Dr Jan C. Oostervilk,*** *Groningen,NL*

**14:30 Update CHEK2: the Dutch experience**
Dr Muriel Adank, Amsterdam, NL

**15:00 How to deal with cancer gene panels**
Dr Ian Frayling, Cardiff, UK

**15:30 Informing family members in hereditary tumour syndromes** Fred H. Menko, Amsterdam, NL

**15:15 De novo loss of function mutations in USP9X cause a female specific recognizable syndrome with developmental delay and distinct congenital malformations** Margot R.F. Reijnders, Nijmegan, NL

**15:30 Understanding mechanisms behind renal cancer development** Elaine Dunlop, Cardiff, UK

**15:45 NF1 genetic testing: where have we got to?** Susan Huson, Manchester, UK

**15:45 Technological innovation in hereditary cancer risk assessment** A Kulkarni, London, UK

**16:00** **SESSION VII: Award ceremony Dutch and UK presentations** **Assembly Room**

**Professor Ruth Newbury-Ecob**, UK & **Dr Frederik Hes**, NL

**16:30** **Main Conference Close**