UK Clinical Genomics 2021

ACGS Association for Clinical Genomic Science

BSGM The British Society for Genetic Medicine

Day 1: Tuesday 19th January 2021

09.50-10.00 "Welcome address" **Eamonn Sheridan**, (*Chair BSGM)*

Delivering genomic medicine in the UK

10.00 – 10.20 "To infinity and beyond – bringing genomic research into the mainstream of healthcare", **Chris Wigley**, (*Genomics England)*

10.20 – 10.40 Sue Hill, *(NHSE)*

10.40 -11.00 "A Flexible, Scalable and Evidence-Based Trio Whole Exome Service for Severe Developmental Disorders in Scotland", **David Fitzpatrick**, *(University of Edinburgh)*

Applications of genomics in human disease

11.00-11.30 "Where next for undiagnosed patients: burden analyses in the DDD study", **Matthew Hurles**, *(Wellcome Sanger Institute)*

11.30-12.00 James Ware, *(Imperial College)*

12.00-13.00 Lunch (Sponsors)

Accepted abstracts

13.00 – 13.15 PRIM1 Deficiency Causes a Distinctive Primordial Dwarfism Syndrome, **Lukas Tamayo Orrego**, (*MRC Human Genetics Unit, Institute of Genetics and Molecular Medicine, University of Edinburgh*)

13.15 – 13.30 NHSE&I R14 Rapid Exome Sequencing Service: Improving management of rare genetic diseases for acutely unwell children, **Hannah Robinson**, (*Exeter Genomics Laboratory, Royal Devon & Exeter NHS Foundation Trust*)

13.30 – 13.45 Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine, **Victor Faundes,** (*Division of Evolution & Genomic Sciences, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester*)

13.45 – 14.00 Isoform-specific variants in the FGF13 gene cause an X-linked early infantile epileptic encephalopathy, **Andrew Fry**, *(Institute of Medical Genetics, University Hospital of Wales)*

14.00 – 14.15 Splicing branchpoint variants contribute to rare disease in the 100,000 Genomes Project, **Alexander Blakes,** *(Manchester Centre for Genomic Medicine, University of Manchester)*

14.15- 14.30 cGAS-mediated induction of type I interferon due to inborn errors of histone pre-mRNA processing, **Yanick Crow**, *(Centre for Genomic and Experimental Medicine, MRC Institute of Genetics and Molecular Medicine, University of Edinburgh)*

14.30-15.00 Break (Genetic Alliance, Swan)

Lightning talks

15.00 – 15.05 Risk assessment and management of rare diseases in the covid19 era: A rapid response, **Elizbeth Forsythe,** (*Clinical Genetics Unit, Great Ormond Street Hospital NHS foundation trust)*

15.05 – 15.10 No association between SCN9A and monogenic human epilepsy disorders, **James Fasham,** *(RILD Wellcome Wolfson Centre, University of Exeter Medical School, Royal Devon & Exeter NHS Foundation Trust)*

15.10 – 15.15 A candidate modifier and autosomal recessive cause of Brugada syndrome that may alter the circadian expression of SCN5A, **Lydia M Seed,** *(Department of Medical Genetics, University of Cambridge)*

15.15-15.20 Functional Analysis of Congenital Stationary Night Blindness Mutations for Therapeutic Intervention, **Tal T Sadeh,** *(Division of Evolution and Genomic Sciences, Faculty of Biology, Medicine and Health, University of Manchester, Manchester)*

15.20 -15.25 Evaluation of prenatal and postnatal genetic testing in pregnancies with fetal anomaly to inform development of a fetal exome service, **Jessica Woods,** *(Leeds Teaching Hospitals NHS Trust)*

15.25-15.30 An Amish founder variant within Smad nuclear interacting protein-1 (SNIP1) associated with an autosomal recessive neurodevelopmental disorder, **Lettie Rawlins,** (*RILD Wellcome Wolfson Centre, University of Exeter Medical School, Royal Devon & Exeter NHS Foundation Trust)*

15.30 – 15.35 Long term evaluation and treatment of women referred to a Breast Cancer Family History Risk and Prevention Clinic (Manchester UK), **Gareth Evans**, *(Nightingale/Prevent Breast Cancer Centre, Wythenshawe Hospital, Manchester University NHS Foundation Trust)*

15.35 -15.40 Dramatic response of metastatic cutaneous angiosarcoma to an immune checkpoint inhibitor in a patient with xeroderma pigmentosum, **Sophie Momen**

15.40 – 15.45 Whole genome sequencing of retinoblastoma reveals the diversity of rearrangements disrupting RB1 and uncovers a treatment related mutational signature, **Helen Davies,** *(Academic Dept of Medical Genetics, University of Cambridge)*

15.45 – 15.50 Clinical Utility and Validity of ctDNA in Management of High Grade Serous Ovarian Cancer, **Djemilah Gordon,** *(West Midlands Regional Genetics Laboratory, Birmingham Women's Hospital)*

16.00-17.00 The BSGM-ACGS Lecture 2021

"MicroRNA biogenesis and tumor susceptibility**", William Foulkes,** *(McGill Centre for Translational Research)*

Day 2: Wednesday 20th January 2021

09.50-10.00 "Welcome address" **Simon Ramsden**, (*Chair ACGS)*

Approaches to treatment for genetic disease

10.00-10.30 "Prenatal cell and gene therapy to treat genetic disease before birth", **Anna David**, *(UCL)*

10.30-10.50 "Disrupting FGFR3 signalling with synthetic C-natriuretic peptide: The A-Z guide of clinical trials in achondroplasia" **Melita Irving**, *(Guy's and St Thomas' NHS Trust)*

10.50-11.20 "Rapid diagnostics to make drug prescription safer and more effective", **William Newman**, University of Manchester

Changing practice in UK clinical genomics

11.20-11.30 "Development of BSGM prenatal guidance and revision of the BSGM genetic testing of children guidance" **Alison Hall and Rachel Hart,** *(BSGM)*

11.30-11.50 "Supporting our colleagues to mainstream genomic medicine", **Amanda Pichini**, *(Heath Education England)*

11.50-12.05 "COVID-19 audit", **Flora Joseph,** *(Cwnselydd Geneteg)*

12.05-13.05 Lunch (NIHR early career researchers meeting, Sponsors)

Accepted abstracts

13.05 -13.20 Missense3D-DB: an atom-based analysis and web catalogue of 4M human protein-coding genetic variation, **Alessia David**, *(Centre for Structural and System Biology, Department of Life Sciences, Imperial College London)*

13.20-13.35 A compendium of mutational signatures of environmental agents, **Xueqing Zou**

13.35-13.50 The Clinical Actionability of Cancer Whole Genome Sequencing (WGS); maximising the value of WGS and assessing the clinical utility, **Kirsty Russell,** *(Bristol Genetics Laboratory, SWGLH)*

13.50-14.05 Heterozygous lamin B1 and lamin B2 variants cause primary microcephaly and define a novel laminopathy, **Andrew Jackson**

Lightning talks

14.05-14.10 dasper: Detection of aberrant splicing events from RNA-sequencing data, **David Zhang**, (*Institute of Child Health, University College London (UCL))*

14.10 -14.15 Splicing noise is detectable across human tissues and modelling its characteristics is likely to improve the detection of pathogenic splicing within patient-derived samples, **Sonia García Ruiz,** (*Department of Genetics and Genomic Medicine Research & Teaching, UCL Great Ormond Street Institute of Child Health)*

14.15-14.20 Detection of Mosaic Chromosomal Alterations in Children with Developmental Disorders, **Ruth Eberhardt,** (*Wellcome Sanger Institute, Wellcome Genome Campus)*

14.20-14.25 Diagnostic yield of Next Generation Sequencing cardiac gene panel testing in patients with inherited cardiac conditions in the Republic of Ireland, **Jane L Murphy,** (*School of Medicine, University College Dublin)*

14.25-14.30 Molecular diagnosis of patients with suspected primary ciliopathies recruited to the 100,000 Genomes Project, **Sunaya Best**, (*Division of Molecular Medicine, Leeds Institute of Medical Research at St. James, University of Leeds)*

14.30-14.35 TierUp: Automated reanalysis of undiagnosed rare disease patients, **Joo Wook Ahn**, *(East Midlands and East of England NHS Genomic Laboratory Hub)*

14.35-14.40 DECIPHER (https://decipher.sanger.ac.uk) – Enabling the sharing and interpretation of rare disease genomic variation and clinical phenotypes, **Julia Foreman**, (*Wellcome Sanger Institute, Wellcome Genome Campus)*

14.40-14.45 Evaluating variants classified as pathogenic in ClinVar in the DDD Study, **Caroline Wright,** (*Institute of Biomedical and Clinical Science, University of Exeter Medical School)*

15.00-15.30 Break (Unique)

The interpretation of DNA variants

15.30-15.50 “Short tandem repeat expansions in the 100,000 Genomes Project”, **Arianna Tucci**

15.50-16.10 Dominic McMullan, *(West Midlands, Oxford and Wessex Genomic Laboratory Hub)*

16.10-16.30 “ACGS guidelines for reporting variants of uncertain significance” **Sian Ellard**, *(South West Genomic Laboratory Hub)*

16.30-17.00 “ClinGen Sequence Variant Interpretation Work Group recommendations for ACMG/AMP guideline specification”, **Steven Harrison**, *(ClinGen)*

17.00 BSGM AGM