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)


13.30 – 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, Elizabeth 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)
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