Genomics England Research Conference Agenda

4 November 2019

08:00 *Room: Entrance Hall*

Registration opens

09:00–09:15 *Location: Great Hall*

Welcome

Chris Wigley (CEO, Genomics England)

09:15–09:20 *Location: Great Hall*

Opening address

Matt Hancock (Secretary of State for Health and Social Care)

09:20–10:05 *Location: Great Hall*

Keynote plenary: Drug Development in the Era of Precision Medicine

Professor David Goldstein (Director of the Institute for Genomic Medicine, Columbia University)

10:05–10:20 *Location: Great Hall*

Keynote sponsor

Hans Cobben (CEO, Bluebee)

10:20–10:50 Coffee/tea break (with posters) *Location: Library & Lecture Hall*

Parallel session 1: Beyond the Coding Regions

Chairs: Professor Tim Hubbard (Genomics England) and Dr. Freya Boardman-Pretty (Genomics England)

Location: Robert Perks

10:50–11:05

Investigating the Microbiome of the 100,000 Genomes Colorectal Cancer Cohort

Dr. Henry Wood (University of Leeds)

11:05–11:20

Filtering Artefacts in Somatic Single Nucleotide Variant Calling Using a Panel of Normals

Dr. Boris Noyvert (University of Birmingham)

11:20–11:35

Real World Experience of Whole Genome Sequencing (WGS) in Haematological Malignancies

Dr. Angela Hamblin (Oxford Genomic Medicine Centre)

11:35–11:45

Xdrop™ - Targeted Sequencing into the Dark and Unknown

Dr. Marie Mikkelsen (Samplix)

Parallel session 2: Diagnostic Discovery

Chair: Dr. Anna Need (Genomics England)

Location: George Thomas

10:50–12.25

Discussion of how to identify novel potential diagnoses in the Research Environment with case studies and examples

Professor Krishna Chatterjee (Institute of Metabolic Science, University of Cambridge)

Dr. Jana Vandrovcova (UCL Queen Square Institute of Neurology Department of Neurodegenerative Disease)

Dr. Jenny Taylor (Wellcome Centre for Human Genetics, Oxford)

Dr. Alistair Pagnamenta (Wellcome Centre for Human Genetics, Oxford)

Dr. Hywel Williams (Cardiff University)

Dr. Guillermo del Angel (Alexion)

12:25–12:30

How Tos & low-hanging fruit: Inspire and encourage Discovery Forum and GeCIP researchers to do diagnostic discovery work

Dr. Anna Need (Genomics England)

Parallel session 3: Precision Clinical Studies in Cancer

Chairs: Dr. Maria Antonietta Cerone (Cancer Research UK) and Dr. Lea Lahnstein (Genomics England)

Location: Great Hall

10:50–11:05

Patient Stratification in Chronic Lymphocytic Leukaemia - Using Whole Genome Sequencing and Machine Learning to Predict Patient Outcomes in Cancer

Dr. Kate Ridout (University of Oxford)

11:00–11:25

The frontline of molecular tumour characterisation for precision medicine in cancer: How Indivumed has developed its IndivuType, a truly multiomics and global cancer database for R&D

Dr. Hartmut Juhl (Indivumed)

11:25–11:40

Identifying fusion protein variants in 100,000 Genomes Project cancer participants for clinical trials

Dr. John Bridgewater (GeCIP PI)

11:40–11:50

Genomics England and precision clinical studies: short introduction and discussion

Frank Nankivell (Genomics England)

Bioinformatics drop-in clinic

Location: Library

Chair: Dr. Loukas Moutsianas (Genomics England)

Drop-in clinic for questions on bioinformatics and tools for working in the Genomics England Research Environment

Parallel session 1: Beyond the Coding Regions

Location: Robert Perks

11:45–11:55

Genome-Wide Map of Selective Constraint from 28,000 Deep Whole Genomes Informs New Model for Non-Coding Pathogenicity

Patrick Short (Wellcome Sanger Institute)

11:55–12:15

Whole Genome Sequencing of Primary Immunodeficiency Reveals a Role for Common and Rare Variants in Coding and Non-Coding Regions

Professor Ken Smith (University of Cambridge)

12:15–12:30

Functional and *In-Silico* Interrogation of Rare Genomic Variants Impacting RNA Splicing for the Diagnosis of Genomic Disorders

Dr. Jamie Ellingford (University of Manchester)

Parallel session 2: Diagnostic Discovery

Location: George Thomas

Parallel session 3: Precision Clinical Studies

Location: Great Hall

11:50–12:05

Germline Pharmacogenomics: Analysis and feedback of variants to avoid drug toxicity

Dr. Ellen McDonagh (Genomics England)

12:05–12:20

Validity of Whole Genomes Sequencing Results in Neoplasms with a High Tumour Mutational Burden in Precision Medicine

Dr. Marc Ooft (King's College Hospital)

12:20–12:30

General discussion on precision studies in cancer and the role of whole genome sequencing

Dr. Maria Antonietta Cerone (Cancer Research UK) & Dr. Lea Lahnstein (Genomics England)

Bioinformatics drop-in clinic

Location: Library

Chair: Dr. Loukas Moutsianas (Genomics England)

Drop-in clinic for questions on bioinformatics and tools for working in the Genomics England Research Environment

12:30–13:20 **Lunch (with posters)** *Location: Library & Lecture Hall*

Connecting patients, participants and researchers as partners

Location: Great Hall

Chair: Vivienne Parry (Genomics England)

13:20–13:35

1. A parent's patient journey

Emma Walters (100,000 Genomes Project participant)

2. Recent PPI activity between a patients' group and the relevant GeCIP domain

Fiona Copeland (GeCIP PPI representative)

3. Building a multidisciplinary community for better research – where the future might lie

Jillian Hastings Ward (Chair of the Genomics England National Participant Panel)

13:35–14:20

Panel discussion and open Q&A

Professor Claire Shovlin (Respiratory GeCIP domain lead)

Dr David Church (Endometrial Cancer GeCIP domain lead)

Fiona Copeland (GeCIP PPI representative)

Helen White (Genomics England National Participant Panel member)

Parallel session 1: Cancer Landscapes

Chair: Tahrima Rahim (Genomics England)
Location: Robert Perks

14:25–14:40
The Genetic Immune Landscape of Colorectal Cancers
Dr. Eszter Lakatos *(Queen Mary University of London)*

14:40–14:55
Assessing the Mutational Landscape of Epithelial Ovarian Cancer Using Whole Genome Sequencing Data from the 100,000 Genomes Project
Dr. Mark Evans *(University of Sheffield)*

14:55–15:10
Characterisation of the Somatic Landscape of Adult Glioma
Dr. Ben Kinnersley *(Institute of Cancer Research)*

15:10–15:25
Identifying Clinically Actionable Genetic Abnormalities in Paediatric Acute Lymphoblastic Leukaemia
Professor Anthony Moorman *(Newcastle University)*

Parallel session 2: Diagnostic Discovery - Research Talks

Chair: Dr. Anna Need (Genomics England)
Location: George Thomas

14:25–14:40
Interrogation of the 100,000 Genomes Project Ophthalmic Disease Cohort Reveals Novel Genes, New Associations and Previously Undetectable Mutations
Dr. Gavin Arno *(University College London)*

14:40–14:55
From the Rare Disease Pilot to the Main Program: How Many of the Newly Discovered Causal Variants and Genes can be Identified in the 100,000 Genomes Project?
Dr. Karyn Megy *(University of Cambridge – NIHR BioResource)*

14:55–15:10
Diagnosis of Rare Disease Using Whole Genome Sequencing in the 100,000 Genomes Project: Experience of the First 2000 Cases
Dr. Andrea Haworth *(Congenica)*

15:10–15:25
Increasing the diagnostic yield for craniosynostosis inside Genomics England by systematic biased "digging"
Dr. Eduardo Calpena *(University of Oxford)*

Parallel session 3: New Technologies & the Genomics England Research Environment

Chair: Dr. James Holman (Genomics England)
Location: Great Hall

14:25–14:30
Introduction: Overview of work being done in the Research Environment
Dr. James Holman *(Genomics England)*

14:30–14:50
Lightning talks explaining the commercial/academic utility of the Research Environment
Dr. Helen Griffin *(Newcastle University)*
Dr. Mark Collins *(Helomics)*
Dr. Salih Tuna *(NIHR BioResource, Cambridge University Hospitals)*
Dr. Amy Hawarden *(University of Manchester)*

14:50–15:05
Overview of News, Tools and Features
Georgia Chan, Dr. Chris Odhams *and* ***Dr. Loukas Moutsianas*** *(Genomics England)*

15:05–15:15
UK Health Data Research Alliance: Implementation through trusted research environments
Professor Tim Hubbard *(Genomics England)*

15:15–15:25
Q&A
Dr. James Holman *& all speakers*

15:25–15:55 **Coffee/tea break (with posters)** *Location: Library & Lecture Hall*

Parallel session 1: Patient Data Ecosystems

Chair: Professor Tim Hubbard (Genomics England)
Location: Robert Perks

15:55–16:10
What Can We Learn About Secondary Findings from the 100,000 Genomes Project to Inform Policy and Practice? Findings from a Workshop with Patients, Clinicians, Policymakers, and Researchers
Dr. Saskia Sanderson (University College London)

16:10–16:25
Assessing the Diagnostic Accuracy of Quantitative Phenotypic Risk Scores Based on Electronic Health Care Records for Patients with Rare Genetic Disorders
Dr. Stefanie Mueller (Institute for Health Informatics, University College London)

16:25–16:40
Generating Health Economic Evidence for Genomic Medicine: Opportunities Arising from the 100,000 Genomes Project
Patrick Fahr (University of Oxford)

Parallel session 2: Collaborating on Research

Chair: Vivienne Parry (Genomics England)
Location: George Thomas

15:55–16:05
Introduction
Vivienne Parry (Genomics England)

16:05–16:40
Interactive session on expectations and joint problem solving in groups

Parallel session 3: Diagnostic Discovery – Research Talks

Chair: Dr. Anna Need
Location: Great Hall

15:55–16:10
The 100,000 Genomes Project: Moorfields Eye Hospital NHS Foundation Trust Experience in Uncovering Pathogenic Variants in Inherited Optic Neuropathies
Dr. Neringa Jurkute (UCL Institute of Ophthalmology and Moorfields Eye Hospital NHS Foundation Trust)

16:10–16:25
Bronchiectasis: Developing Early Insights Into An Old Disease
Dr. Helen Griffin (Newcastle University)

16:25–16:40
Whole Genome Sequencing Through the 100,000 Genomes Project Identifies a Novel Cause of Pulmonary Arteriovenous Malformations Associated with Hereditary Haemorrhagic Telangiectasia
Professor Claire Shovlin (Imperial College London)

Research Environment User & Bioinformatics Panel

Chair: Dr. Loukas Moutsianas
Location: Library

15:55–16:40
Research Environment users and Genomics England experts discuss Slido questions from the Research Environment session and open questions from other users
Dr. Chris Odhams (Genomics England)
Dr. Georgia Chan (Genomics England)
Dr. Alona Sosinsky (Genomics England)
Dr. Mark Collins (Helomics)
Dr. Hywel Williams (Cardiff University)
Dr. Patrick Short (SanoGenetics)

Closing plenary

Room: Great Hall

16:50–17:00
Recap of the day

17:00–17:15
The National Genomic Healthcare Strategy
Professor Sir John Bell (Genomics England Board Member)

17:15–17:30
Genomics England – the future: Update Towards 5 Million Genome Analyses
Professor Sir Mark Caulfield (Chief Scientist, Genomics England)

17:30–17:35
Towards 5 Million
UCB Industry Cohort

17:35–17:50
Genomics England – the future: Closing remarks by Chris Wigley, CEO of Genomics England

17:50–19:30 **Drinks reception** *Room: Library & Lecture Hall*

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