Immunogenomics of Disease: Accelerating to Patient Benefit
5-7 February 2019

Wellcome Genome Campus,
Hinxton, Cambridge, UK

Conference Programme

Tuesday 5 February 2019

12:00-13:00  Registration with lunch

13:00-13:10  Welcome and introduction
Programme Committee: Sophie Hambleton, Newcastle University and Great North Children’s Hospital, UK

13:10-14:10  Keynote 1
Chair: Sophie Hambleton, Newcastle University and Great North Children’s Hospital, UK

Can the study of monogenic diseases of the immune system contribute to treat better more common and complex diseases
Alain Fischer
Imagine Institute, Collège de France, France

14:10-15:25  Session 1: High-throughput approaches to understanding immune cell function I
Chair: Julian Knight, University of Oxford, UK

14:10  Single cell genomics: using finer lenses to unravel features of human immunity
Chloé Villani
Massachusetts General Hospital, USA

14:40  Single-cell genomics: a new understanding of the human immune system in health and disease
Monika Kowalczyk
Celsius Therapeutics, USA
15:10  Sharing information between related diseases using Bayesian joint fine mapping increases accuracy and identifies novel associations in six immune mediated diseases
   Chris Wallace
   University of Cambridge, UK

15:25-16:00  Afternoon tea

16:00-17:15  Session 2: High-throughput approaches to understanding immune cell function II
   Chair: Carl Anderson, Wellcome Sanger Institute, UK

   16:00  Deciphering human immune landscape for precision medicine
          Derya Unutmaz
          Jackson Laboratory, USA

   16:30  Generating human cell-type-specific protein interaction networks to functionally interpret genetic data
          Kasper Lage
          MIT, USA

   17:00  Disease mechanism and genetic subtyping revealed through informed dimension reduction of the immune-mediated disease genome
          Oliver Burren
          University of Cambridge, UK

17:15-17:40  Lightning talks for poster session I
   Chair: Carl Anderson, Wellcome Sanger Institute, UK

17:40-19:10  Poster session I (odd numbers) with drinks reception

19:10  Dinner
Wednesday 6 February 2019

09:00-10:30  Session 3: From genetic discovery to patient benefit I  
Chair: Julian Knight, University of Oxford, UK

09:00  Using human genetics to transform drug discovery  
Jeffrey Barrett  
Genomics Plc, UK

09:30  The 11q13.5 immune risk locus contains a distal enhancer required for  
Treg-mediated suppression of gut inflammation  
Rahul Roychoudhuri  
Babraham Institute, UK

10:00  PICCOLO: a new tool enabling testing colocalization between eQTL  
and GWAS hits in the absence of full summary statistics  
Jorge Esparza Gordillo  
GlaxoSmithKline, UK

10:15  Germline TET2 loss-of-function causes childhood immunodeficiency  
with lymphoma predisposition  
Jarmila Spegarova  
Newcastle University, UK

10:30-11:00  Morning coffee

11:00-12:30  Session 4: From genetic discovery to patient benefit II  
Chair: Nadine Cerf-Bensussan, Inserm, France

11:00  Adapting AI to rare diseases  
Anita Burgun  
Imagine, France

11:30  Gene therapy for inherited disease of the hematopoietic systems: advances and future challenges  
Marina Cavazzana  
Imagine, France

12:00  Translating osteoarthritis GWAS signals into therapeutic targets  
Linda McCarthy  
GlaxoSmithKline, UK

12:15  Using in vivo eQTL interactions to identify the regulatory drivers of  
variation in the transcriptomic response to sepsis  
Emma Davenport  
Wellcome Sanger Institute, UK

12:30-14:00  Lunch
14:00-15:30  **Session 5: Immune-repertoire sequencing as a tool to understand adaptive immunity**  
*Chair: Sophie Hambleton, Newcastle University and Great North Children’s Hospital, UK*

14:00  **Monitoring the dynamics in B cell responses following infection and vaccination using immune repertoire sequencing**  
*Velislava Petrova*  
*Wellcome Sanger Institute, UK*

14:30  **Adaptive immune receptor repertoires in coeliac disease**  
*Ludvig Sollid*  
*University of Oslo, Norway*

15:00  **Genomic dissection of variation in patient response to checkpoint inhibitors**  
*Benjamin Fairfax*  
*University of Oxford, UK*

15:15  **Elucidating the role of immunoglobulin heavy chain locus polymorphism on antibody diversity and function**  
*Oscar Rodriguez*  
*Icahn School of Medicine at Mount Sinai, USA*

15:30-16:00  **Afternoon tea**

16:00-17:30  **Session 6: Immunophenomics**  
*Chair: Carl Anderson, Wellcome Sanger Institute, UK*

16:00  **The human phenotype ontology: a semantic framework for phenotype driven translational research and genomic diagnostics**  
*Peter Robinson*  
*The Jackson Laboratory for Genomic Medicine, USA*

16:30  **Insights from extreme immune phenotypes**  
*Sophie Hambleton*  
*Newcastle University and Great North Children’s Hospital, UK*

17:00  **Early onset autoimmunity and CMV susceptibility due to a hypomorphic IL-2 receptor beta mutation**  
*Florian Gothe*  
*Newcastle University, UK*

17:15  **Longitudinal serological measures of common infection**  
*Ruth Mitchell*  
*University of Bristol, UK*
17:30-18:00  **Lightning talks for poster session 2**  
*Chair: Carl Anderson, Wellcome Sanger Institute, UK*

18:00-19:30  **Poster session 2 (even numbers) and drinks reception**

19:30  **Conference dinner**

---

**Thursday 7 February 2019**

08:30-09:30  **Keynote 2**  
*Chair: Julian Knight, University of Oxford, UK*

**Translating immunology into new targets for IBD**  
*Fiona Powrie*  
*The Kennedy Institute of Rheumatology, UK*

09:30-10:30  **Session 7: The population genetics of the immune-system**  
*Chair: Sophie Hambleton, Newcastle University and Great North Children’s Hospital, UK*

09:30  Progression from benign to pathogenic autoantibody by somatic mutation in a rogue clone: single cell RNA and DNA sequencing  
*Chris Goodnow*  
*Garvan Institute of Medical Research, Australia*

10:00  Human immune system variation and development early in life  
*Petter Brodin*  
*Karolinska Institute, Sweden*

10:30  Genetic architecture of adaptive immune system identifies key immune regulators  
*An Goris*  
*KU Leuven, Belgium*

10:45-11:15  **Morning coffee**

11:15-12:45  **Session 8: Cellular resolution of complex diseases**  
*Chair: Nadine Cerf-Bensussan, Inserm, France*

11:15  Single cell sequencing and Crohn’s disease heterogeneity  
*Judy Cho*  
*Icahn School of Medicine at Mount Sinai, USA*
11:45  Chromatin contacts and transcriptomics in CD4+ T-cells reveal genes implicated in rheumatoid arthritis
   Vasanthi Priyadarshini Gaddi
   University of Manchester, UK

12:00  Cell type resolution of cis-eQTL in GTEx tissues
   Sarah Kim Hellmuth
   New York Genome Center, USA

12:15  Contribution of a risk allele and sex hormones to a lupus phenotype
   Betty Diamond
   The Feinstein Institute for Medical Research, USA

12:45-12:55  Closing remarks
   Programme Committee: Julian Knight, University of Oxford, UK

12:55-13:10  Take away lunch

13:10  Coaches depart to Cambridge City Centre via Train Station, and Heathrow Airport via Stansted Airport