

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 I**
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 I: 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