



The Genomics of Common Diseases 6 – 9 September 2017

**Wellcome Genome Campus
Hinxton, Cambridge, UK**

Conference Programme

Wednesday, 6 September

13:00 – 14:45 Registration with lunch

14:45 – 15:00 Welcome and Introduction by Orli Bahcall & Nicole Soranzo

15:00 – 15:45 **Keynote Lecture**

Chair: Orli Bahcall Nature, USA

Immunogenomics at high resolution

Sarah Teichmann

Wellcome Trust Sanger Institute, UK

15:45 – 16:15 Afternoon Tea

16:15 – 18:00 **Session 1: Identifying novel genes and pathways**

Chair: Teri Manolio National Human Genome Research Institute, USA

16:15 Title tbc

John Danesh

UKBB Cambridge, UK

16:40 Realising the power of large prospective biobanks in diverse populations

Zhengming Chen

University of Oxford, UK

17:05 The Genetics of Obesity: using alternative approaches to target new biology

Ruth Loos

Icahn School of Medicine at Mount Sinai, USA

17:30 Expanding the spectrum of type 2 diabetes risk alleles through genome-wide association study imputed up to Haplotype Reference Consortium panel

Anubha Mahajan

WTCHG, University of Oxford, UK

17:45 Copy number variation and breast cancer risk
Joe Dennis
University of Cambridge, UK

18:00 – 19:00 Drinks Reception

19:00 – 21:00 Dinner

Thursday, 7 September

09:00 – 10:45 **Session 2: Regulatory variation of complex human diseases**
Chair: Kyle Vogan Nature Genetics, USA

09:00 Speaker tbc

09:25 Induced pluripotent stem cells as models for human diseases
Helena Kilpinen
University College London, UK

09:50 Long-distance relationships and gene regulation in development and disease
Francois Spitz
Institut Pasteur, France

10:15 High-resolution genetic mapping of causal regulatory interactions
Daniel Gaffney
Wellcome Trust Sanger Institute, UK

10:30 Transcriptome-wide association studies are vulnerable to false positives due to co-regulation
Michael Wainberg
Stanford University, USA

10:45 – 11:25 Morning Coffee

11:25 – 13:10 **Session 3: Advances in large-scale functional annotation**
Chair: Nicole Soranzo Wellcome Trust Sanger Institute, UK

11:25 Non-coding mutations in the 3-Dimensional Genome
Jim Hughes
University of Oxford, UK

11:50 Insight into blood production from functional follow up of human genetic studies
Vijay Sankaran
Broad Institute, USA

- 12:15 Oncogene activation by structural variant-mediated alteration of the cis-regulatory landscape
Joachim Weischenfeldt
BRIC, Denmark
- 12:40 Large-scale protein-protein interaction experiments of schizophrenia risk genes in human neurons coalesce GWAS loci into unexpected pathways
Kasper Lage
Broad Institute of MIT and Harvard, USA
- 12:55 Investigating chromatin interactions at Type 2 Diabetes associated loci in human –cells
Vibe Nylander
University of Oxford, UK

13:10 – 14:40 Lunch

14:40 – 16:25 **Session 4: Innovative approaches to complex trait analysis**

Chair: Orli Bahcall Nature, USA

- 14:40 Inference on natural selection and genetic (co)variation from large GWAS data
Peter Visscher
University of Queensland, Australia
- 15:05 Identifying disease-relevant cell types using GWAS data
Hilary Finucane
Harvard Chan School, USA
- 15:30 Imaging genetics and fine-scale population structure in the UK Biobank
Jonathan Marchini
University of Oxford, UK
- 15:55 Re-evaluation of SNP Heritability in Complex Human Traits
David Balding
UCL and Melbourne, UK
- 16:10 Genome-wide association study of social genetic effects
Amelie Baud
EMBL-EBI, UK

16:25 – 17:05 Afternoon Tea

17:05 – 17:50 **Keynote Lecture**

Chair: Sekar Kathiresan Massachusetts General Hospital, USA

Using human genetics & genomics to unravel causal mechanisms for diabetes

Anna Gloyn
University of Oxford, UK

17:50 – 19:20 **Poster session 1 with drinks reception**

19.20 – 21:00 Dinner

Friday, 8 September

09:00 – 10:45 **Session 5: Using human genetics to inform drug development**

Chair: Sekar Kathiresan Massachusetts General Hospital, USA

- 09:00 Using Human Genetics for Drug Development: the Industry Perspective
*Caroline Fox
Merck, USA*
- 09:25 Drug Development in the Era of Human Genetically Validated Targets: Case Studies at Amgen
*Simon Jackson
Amgen, USA*
- 09:50 The role of genetic evidence in drug discovery and development: past, present and future
*Robert Scott
GlaxoSmithKline, USA*
- 10:15 A novel LDL-lowering missense variant in B4GALT1 identifies novel biological connection between protein glycosylation and cardiovascular risk factors in human
*May Montasser
University of Maryland, USA*
- 10:30 Meta-analysis of genome-wide association studies in four systemic autoimmune diseases to identify shared genetic etiologies
*Marialbert Acosta Herrera
Instituto de Parasitología y Biomedicina López-Neyra, Spain*

10:45 – 11:25 Morning Coffee

11:25 – 13:10 **Session 6: Bringing genomics into clinical use**

Chair: Teri Manolio National Human Genome Research Institute, USA

- 11:25 Germline genetic testing for cancer: risk prediction to therapy
*Susan Domchek
University of Pennsylvania, USA*
- 11:50 Full genome analysis for genetic disease diagnosis
*Pui Kwok
UCSF, USA*

12:15 Integrating Omics with Clinical Outcomes

Claudia Langenberg
MRC Cambridge, UK

12:40 Gaining precision on statin intolerance with genomics

Marie Pierre Dube
Montreal Heart Institute, Canada

12:55 Estimating genetic parameters for common diseases from large-scale population health data sets

Christopher DeBoever
Stanford University, USA

13:10 – 14:40 Lunch

14:40 – 16:15 **Session 6: continued**

14:40 Title tbc

Nancy Cox
Vanderbilt University, USA

15:05 Precision oncology for acute myeloid leukemia using a knowledge bank approach

Moritz Gerstung
EMBL-EBI, UK

15:30 Coding and noncoding cancer mutations

Nuria Lopez-Bigas
Institute for Research in Medicine, Spain

15:55 Generation Scotland: Scottish Family Health Study: Linkage to Electronic Health Records

Caroline Hayward
University of Edinburgh, UK

15:10 Monogenic and polygenic predictors of extreme dyslipidemia from whole genome sequencing in 8,394 white and black NHLBI TOPMed participants

Gina Peloso
Boston University, USA

16:15 – 16:50 Afternoon Tea

16:50 – 17:35 **Keynote Lecture**

Chair: Nicole Soranzo Wellcome Trust Sanger Institute, UK

Common diseases, related physiologic functions and the effects of common and rare variants on both

Kari Stefansson
deCODE genetics, USA

17:35 – 19:05 **Poster session 2 with drinks reception**

19:05 – 21:00 Conference Dinner

Saturday, 9 September

08:45 Coaches to Cambridge and Heathrow Airport via Stansted Airport depart

Optional extra day (registration required)

**Big Data Analytics for Genetics in Personalised Medicine
9 September 2017**

**Wellcome Genome Campus
Hinxton, Cambridge, UK**

Conference Programme

Saturday, 9 September

09:00 – 09:30 Arrival coffee

09:30 – 09:35 Welcome and Introduction

09:35 – 10:05 Phenotype-driven genome diagnostics
Peter Robinson
The Jackson Laboratory, USA

10:05 – 10:35 Standards and Approaches to Support Genomic Variant Interpretation
Heidi Rehm
The Broad Institute, USA

10:35 – 11:05 Statistical approach for investigating change in mutational process during cancer growth and development
Kimberly Siegmund
University of Southern California, USA

11:05 – 11:30 Morning coffee

11:30 – 12:00 Mendelian randomization in a data rich environment
George Davey Smith
University of Bristol, UK

12:00 – 12:30 Integrating GWAS, omics, and electronic medical records to dissect disease biology
Haky Im
University of Chicago, USA

12:45 Coaches to Cambridge and Heathrow Airport via Stansted Airport depart