Genome Informatics

17-20 September 2018

Wellcome Genome Campus Conference Centre, Hinxton, Cambridge, UK

Conference Programme

Monday, 17 September

13:00 – 14:50 **Registration with lunch**

14:50 – 15:00 **Welcome and Introductions**
Aaron Quinlan, University of Utah USA

15:00 – 16:20 **Session 1: Data Curation, Integration, and Visualization**
Session chairs: Casey Green & Sarah Teichmann

15:00 Immunogenomics one cell at a time
Sarah Teichmann
Wellcome Sanger Institute, UK

15:20 Flexible and interactive visualization of GFA sequence graphs
Giorgio Gonnella
University of Hamburg, Germany

15:40 Using clustering trees to visualise single-cell RNA-sequencing data
Luke Zappia
Murdoch Children's Research Institute, Australia

16:00 Expression Atlas: exploring gene expression at tissue and single cell level across species and biological conditions
Laura Huerta
EMBL-EBI, UK

16:20 – 17:00 **Afternoon Tea**

17:00 – 18:20 **Session 1 continued**

17:00 Interpreting high-throughput genomic analyses with the het.io knowledgebase
Casey Green
University of Pennsylvania, USA

17:20 Pathogens in Ensembl: Enabling the march against biotic threat
Nishadi De Silva
EMBL-EBI, UK

17:40 Butler enables rapid analysis of thousands of human genomes on the cloud.
Sergei Yakneen
EMBL, Germany

18:00 InterMine: widening integrative data analysis
Rachel Lyne
InterMine, UK

18:20 – 19:00 **Drinks reception**

19:00 **Dinner**

Tuesday, 18 September

09:00 – 09:55 **Keynote Lecture**
Session chair: Melissa Wilson Sayers, Arizona State University, USA

A population genetic view of human chromatin organization
Katie Pollard
Gladstone Institute of Data Science & Biotechnology / UCSF, USA

09:55 – 10:00 Comfort break

10:00 – 11:20 **Session 2: Personal and Medical Genomics**
Session chairs: Sri Kosuri & Kaitlin Samocha

10:00 The Impact of Rare Genetic Variation on Pre-mRNA Splicing
Sri Kosuri
UCLA, USA

10:20 Integration of whole genome, whole exome, and transcriptome sequencing pipelines for comprehensive genomic profiling of 55 pediatric cancer subjects
Patrick Brennan
Nationwide Children's Hospital, USA

10:40 Non-driver somatic alterations confer good prognosis in lung cancer patients
Dennis Wang
NIHR Sheffield BRC, UK

11:00 Resolving tumor heterogeneity at single cell resolution
Shamoni Maheshwari
10x Genomics, USA

11:20 - 11:50 **Morning Coffee**

11:50 – 13:10 **Session 2 continued**

11:50 Evaluating the role of rare variation in children with developmental disorders
Kaitlin Samocha
Wellcome Sanger Institute, UK

12.10 Inference of mutational status, loss of heterozygosity, and clonality in tumor-only data
Hossein Khiabanian
Rutgers University, USA

12.30 Modelling double strand break susceptibility to interrogate structural variation in cancer
Tracy Ballinger
IGMM, UK

12.50 Retrieving Charras genomic tracts from the Uruguayan admixed population
Lucia Spangenberg
Institut Pasteur de Montevideo

13:10 – 14:30 **Lunch**

14:30 – 15:50 **Session 3: Comparative, Evolutionary, Metagenomics**
Session chairs: Mario Caccamo & Ellen Leffler

14:30 Paired sequencing of host and parasite genomes in severe malaria cases
Ellen Leffler
University of Oxford, UK

14:50 PPanGGOLiN: Depicting microbial diversity via a Partitioned Pangenome Graph
Guillaume Gautreau
Genoscope, France

15:10 The role of structural variants in the adaptive radiation of African Cichlids
Luca Penso Dolfin
Earlham Institute, UK

15:30 Coevolution of chromosome changes and gene regulation in ruminants
Marta Farre Belmonte
Royal Veterinary College

15:50 – 16:30 **Afternoon Tea**

16:30 – 17:50 **Session 3 continued**

16:30 Understanding the genetic components controlling apomixis
Mario Caccamo
NIAB, UK

16:50 Querying colored and compacted de Bruijn graphs of thousands of related genomes
Nina Luhmann
University of Warwick

17:10 Genome mining for metabolic gene clusters in yeast
Christopher Pyatt
NCYC - Quadram Institute, UK

17:30 Comparative analysis of hundreds of vertebrate genomes in Ensembl
Carla Cummins
EMBL-EBI, UK

17:50 – 18:15 **Lightning talks**

18:15– 19:30 **Drinks reception and Poster session I** (Odd numbers)

19:30 **Dinner**

Wednesday, 19 September

09:00 – 09:55 **Keynote Lecture**
Session chair: Alicia Oshlack

Understanding variability and systematic bias in highthroughput data
Rafael Irizarry
Dana-Farber Cancer Institute, USA

09:55 – 10:00 Comfort break

10:00 – 11:20 **Session 4: Transcriptomics, Alternative Splicing and Gene Predictions**
Session chairs: Barbara Englehardt & Mark Robinson

10:00 On the analysis of long-read sequencing data for gene expression
Mark Robinson
University of Zurich, Switzerland

10:20 Single-cell isoform RNA sequencing (ScISOr-Seq) across thousands of cells reveals isoforms of cerebellar cell types.
Hagen Tilgner
Weill Cornell Medicine, USA

10:40 Bootstrapping Biology: Quick and easy de novo genome assembly to enable single cell gene expression analysis
Nikka Keivanfar
10x Genomics, USA

11:00 Discrete and continuous differential expression analysis for single-cell RNA-seq data
Koen Van den Berge
Ghent University, Belgium

11:20 - 11:50 **Morning Coffee**

11:50 – 13:10 **Session 4 continued**

11:50 A generative model for single-cell RNA-sequencing
Barbara Englehardt
Princeton University, USA

12.10 TALC: Transcriptome-aware Long Read Correction
Lucile Broseus
CNRS, France

12.30 Constraint for mRNA structure in human synonymous mutations
Jeff Gaither
Nationwide Children's Hospital, USA

12.50 Differential isoform usage in Parkinson's disease
Fiona Dick
University of Bergen, Norway

13:10 – 14:30 **Lunch**

14:30 – 15:50 **Session 5: Epigenetics and non-coding genome**
Session chairs: Jordana Bell & Alexander Suh

14:30 Interpreting variation in the human methylome
Jordana Bell
King's College London, UK

14:50 Delineation and annotation of the human regulatory landscape across 400+ cell types and states
Wouter Meuleman
Altius Institute for Biomedical Sciences, USA

15:10 Tissue-specific enhancer and promoter evolution in mammals
Maša Roller
EMBL-EBI, UK

15:30 Exploratory analysis of retrotransposon activity in the octopus brain
Massimiliano Volpe
Stazione Zoologica Anton Dohrn, Italy

15:50 – 16:30 **Afternoon Tea**

16:30 – 17:50 **Session 5 continued**

16:30 Mind the gap - interrogating the non-coding genome with single-molecule technologies
Alexander Suh
Uppsala University, Sweden

16:50 Identification of genes escaping X-inactivation and the variability of escape across cells, tissues and twin pairs
Antonino Zito
King's College London, UK

17:10 DNA methylation changes as a marker of senescing leaves in Arabidopsis thaliana.
Minerva Susana Trejo Arellano
Swedish University of Agricultural Sciences, Sweden

17:30 Recent evolution of the epigenetic regulatory landscape in human and other primates
Raquel Garcia Perez
Institute of Evolutionary Biology, Spain

17:50 – 18:15 **Lightning talks**

18:15– 19:30 **Drinks reception and Poster session 2** (Even numbers)

19:30 **Conference Dinner, Silver Service**

Thursday, 20 September

09:00 – 10:20 **Session 6: Variant Discovery and Genome Assembly**
Session chairs: Jeff Kidd & Melissa Wilson Sayers

09:00 Sex differences in reference genome affect variant calling and differential expression
Melissa Wilson Sayers
Arizona State University, USA

09:20 Encoding yeast genomic diversity using variation graphs
Prithika Sritharan
Quadram Institute Bioscience, UK

09:40 Genome analysis in a polymorphic moss with large, ancient sex chromosomes
Sarah Carey
University of Florida, USA

10.00 ScaffHiC Genome Scaffolding by Modelling Distributions of Hi-C Paired-end Reads
Zemin Ning
Wellcome Sanger Institute, UK

10:20 - 11:00 **Morning Coffee**

11:00 – 12:20 **Session 6 continued**

11:00 De novo assembly and analysis of a canine genome
Jeff Kidd
University of Michigan, USA

11:20 Pandora variation inference for pangenomes from Nanopore or Illumina data
Rachel Colquhoun
University of Oxford, UK

11:40 Direct measurement of spontaneous structural variation through whole-genome sequencing of three generation human pedigrees
Jonathan Belyeu
University of Utah, USA

12:00 VarTrix is an open-source software tool for assigning variants to individual cells
Ian Fiddes
10x Genomics, USA

12:20 – 12:25 **Closing remarks by the Scientific Programme Committee**

12:20 – 13:30 **Lunch**

13:30 **Close of conference, coaches depart to Cambridge, Stansted and Heathrow airports**