Monday, 19 September

13:00 – 14:50  Registration with lunch

14:50 – 15:00  Welcome and Introductions

15:00 – 16:20  Session 1: Personal and Medical Genomics  
Session chairs: Jessica Chong & Meromit Singer

  15:00  Transforming gene discovery by radically open data sharing  
        Jessica Chong  
        University of Washington, USA

  15:20  Prediction of splice variants and transcript-level effects  
        improves the identification of gene-disrupting variants  
        Konrad Karczewski  
        MGH/Broad Institute, USA

  15:40  Advancing massively parallel reporter assays for interpreting  
        regulatory variants  
        Martin Kircher  
        University of Washington, USA

  16:00  Improving the speed of genome interpretation via network  
        collaboration and knowledge sharing  
        Jennifer Harrow  
        Illumina, UK

16:20 – 17:00  Afternoon Tea

17:00 – 18:20  Session 1 continued

  17:00  A distinct gene module for T cell dysfunction (“exhaustion”)  
        uncoupled from T cell activation  
        Meromit Singer  
        Broad Institute, USA

  17:20  Characterizing genomic responses to cancer immunotherapy  
        Christopher Miller  
        Washington University, USA
17:40 Transcript haplotypes: beyond single variant analysis
William McLaren
EMBL-EBI, UK

18:00 Improving genome analysis using Linked-Reads
Deanna Church
10x Genomics, USA

18:20 – 19:00 Drinks reception
19:00 Dinner

Tuesday, 20 September

09:00 – 10:20 Session 2: Variant Discovery and Genome Assembly
Session chairs: Jared Simpson & Alicia Oshlack

09:00 Analysis methods for nanopore sequencing data
Jared Simpson
Ontario Institute for Cancer Research, USA

09:20 Curation of multiple genome assemblies of the same species and utilisation for reference improvement
Kerstin Howe
Wellcome Trust Sanger Institute, UK

09:40 Processing genomics data at scale with ADAM and Toil
Frank Nothaft
University of California, Berkeley, USA

10:00 RUFUS: Reference free variant detection improves accuracy and sensitivity
Andrew Farrell
University of Utah, USA

10:20 - 11:00 Morning Coffee

11:00 – 12:20 Session 2 continued

11:00 SuperTranscript: a linear sequence assembled for the visualization and analysis of transcriptome data
Alicia Oshlack
Murdoch Childrens Research Institute, Australia

11.20 GCSA2: A scalable approach to indexing population variation graphs
Jouni Sirén
Wellcome Trust Sanger Institute, UK
11.40 Unknown and non-repetitive sequence in the Icelanders genomes
   Birte Kehr
deCODE genetics, Iceland

12:00 Lancet: somatic variant calling using localized colored DeBruijn graphs
   Giuseppe Narzisi
   New York Genome Center, USA

12:20 - 14:00 Lunch

14:00 – 15:20 Session 3: Epigenomics and Non-Coding Genome
   Session chairs: Katie Pollard & Anshul Kundaje

14:00 Decoding enhancer function with machine learning
   Katie Pollard
   University of California, San Francisco, USA

14:20 DeepNuc: A deep learning model that accurately predicts genome-wide nucleosome positioning from ATAC-seq
   Chris Probert
   Stanford University, USA

14:40 Functional categorisation of fly regulatory elements and their regulatory potential by their transcription initiation patterns and nuclear stability of produced RNAs
   Robin Andersson
   University of Copenhagen, Denmark

15:00 Transcription factor expression and its effects on binding site occupancy and motif preference
   Michael Hoffman
   Princess Margaret Cancer Centre, Canada

15:20 – 16:00 Afternoon Tea

16:00 – 17:20 Session 3 continued

16.00 Deep learning transcription factor binding sites and regulatory sequence grammars in diverse cell types and lineages
   Anshul Kundaje
   Stanford University School of Medicine, USA

16:20 Mutational biases drive elevated rates of substitution at regulatory sites across cancer types
   Vera Kaiser
   University of Edinburgh, UK
16:40 Random walk modelling of transcription initiation landscapes reveals three dimensional chromatin organization and spatial transcriptional variability between cell types
Sarah Rennie
University of Copenhagen, Denmark

17:00 Functional genetic screen for enhancer elements in the human genome using CRISPR-Cas9
Rani Elkon
Tel Aviv University, Israel

17:20 – 18:20 Keynote Lecture
Session chair: Aaron Quinlan

Translating the Cancer Genome: computational tools inform therapeutic decisions
Elaine Mardis
Washington University School of Medicine, USA

18:20 – 19:30 Drinks reception and Poster session I (Odd numbers)

19:30 Dinner

Wednesday, 21 September

09:00 – 10:00 Keynote Lecture
Session chair: Daniel MacArthur

New Genome Reference Structures
Richard Durbin
Wellcome Trust Sanger Institute, UK

10:00 – 11:20 Session 4: Data Curation and Visualization
Session chairs: Kim Pruitt & Alex Bateman

10:00 NCBI graphical display tools – Hidden in plain sight
Kim Pruitt
National Institutes of Health, USA

10:20 RNA-seq visualization using Degust
David Powell
Monash University, Australia

10:40 ShinyNGS: Interactive data mining with R and Shiny
Jonathan Manning
University of Edinburgh, UK

11:00 Complex Portal - a unifying protein complex database
Birgit Meldal
EMBL-EBI, UK

11:20 - 11:50 Morning Coffee
Session 4 continued

11:50 The sustainability of literature curation  
Alex Bateman  
EMBL-EBI, UK

12.10 DoCM: a database of curated mutations in cancer rescues clinically relevant mutations  
Benjamin Ainscough  
Washington University School of Medicine, USA

12.30 GeneValidator: Identify problems with protein-coding gene predictions  
Ismail Moghul  
Queen Mary University of London, UK

12.50 GIGGLE: a scalable and fast search engine for large-scale multi-omics data integration  
Ryan Layer  
University of Utah, USA

13:10 – 14:30  
Lunch

Session 5: Transcriptomics, Alternative Splicing and Gene Predictions  
Session chairs: Davis McCarthy & Alexandra-Chloé Villani

14:30 Using single-cell RNA-seq data for inference on gene regulation  
Davis McCarthy  
EMBL-EBI, UK

14:50 Comprehensive transcriptome analysis using synthetic long read sequencing reveals molecular co-association and conservation of distant splicing events  
Hagen Tilgner  
Weill Cornell Medical College, USA

15:10 Spliced synthetic genes as internal controls in RNA sequencing experiments  
Simon Hardwick  
Garvan Institute of Medical Research, Australia

15:30 Fast fusion detection, assembly, and quantification using kallisto  
Pall Melsted  
University of Iceland, Iceland

15:50 – 16:30  
Afternoon Tea

16:30 Dissecting the human immune system in health and disease: insights from unbiased genomics strategies  
Alexandra-Chloé Villani  
Broad Institute, USA
16:50 Meta-analysis of the induced immediate early response across eight CAGE time course datasets
Annalaura Vacca
University of Edinburgh, UK

17:10 A Bayesian model for single cell transcript expression analysis on MERFISH data
Johannes Koester
Centrum Wiskunde & Informatica, Netherlands

17:30 Column subset selection for single-cell RNA-seq clustering
Shannon McCurdy
University of California, Berkeley, USA

17:50 – 19:00 Drinks reception and Poster session II (Even numbers)

19:00 Conference Dinner, Silver Service

Thursday, 22 September

09:00 – 10:20 Session 6: Comparative, Evolutionary, and Metagenomics
Session chairs: Curtis Huttenhower & Alicia Martin

09:00 Functional profiling of metagenomes, metatranscriptomes and strains in the next stages of the NIH Human Microbiome Project
Curtis Huttenhower
Harvard School of Public Health, USA

09:20 Identifying highly constrained protein-coding regions using population-scale genetic variation
James Havrilla
University of Utah, USA

09:40 Horizontal gene transfer from birds to parasitic nematodes? When similarity searches are not enough
Sonja Dunemann
University of Calgary, Canada

10.00 The combined landscape of Denisovan and Neanderthal ancestry in present-day humans
Sriram Sankararaman
University of California, Los Angeles, UK

10:20 - 11:00 Morning Coffee

11:00 – 12:20 Session 6 continued

11:00 Fine-scale identity-by-descent and birth records in Finland provide insights into recent population history
Alicia Martin
Massachusetts General Hospital, USA
11:20 The landscape of human sex-differential transcriptome and its subsequent selection  
*Moran Gershoni*  
*Weizmann Institute of Science, Israel*

11:40 Using phylogenetic instability to predict members of large gene families with xenobiotic functions  
*David Curran*  
*University of Calgary, Canada*

12:00 Updated analysis results from the NIH Human Microbiome Project  
*Owen White*  
*University of Maryland, Institute for Genome Sciences, USA*

12:20 – 12:25 Closing remarks by Janet Kelso and Daniel MacArthur

12:20 – 13:30 **Lunch**

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