

CONFERENCE PROGRAM

DAY 1 THURSDAY 27 SEPTEMBER

11:00	Registration and poster hanging
13:40	Introduction Rebecca Furlong, Genome Medicine, UK
	Informatics workshop Chair: David Dooling
13:45	New algorithms and infrastructure for variant detection at population scale Gabor Marth, Boston College, USA
14:15	Network and pathway analysis of somatic mutations in cancer Ben Raphael, Brown University, USA
14:45	Pathway-based analysis of mutation impact Josh Stuart, University of California Santa Cruz, USA
15:15	Sponsored technical presentation – Strand Aishwarya Narayanan, Application Scientist, Strand
15:15–16:00	Coffee
16:00	Illuminating the genetics of complex human diseases Mike Schatz, Cold Spring Harbor Laboratories, USA
16:30	Accessible, transparent, reproducible analysis with Galaxy James Taylor, Emory University, USA
17:00	Informatics challenge David Dooling, Michael Schatz, James Taylor
18:00	Sponsored technical presentation – DNAnexus Andreas Sundquist, CEO and Co-founder of DNAnexus
18:00–19.30	Poster session 1 and welcome reception (odd-numbered posters to be presented)

CONFERENCE PROGRAM

DAY 2 FRIDAY 28 SEPTEMBER

- 08:00–08:55 **Coffee/breakfast**
Session 1. Inherited disease: beyond the candidate gene approach
 Chair: James Lupski
- 09:00 **Hypothesis-generating clinical genomics research and predictive medicine**
 Leslie Biesecker, National Human Genome Research Institute, USA
- 09:30 **Whole-genome sequencing and disease-gene detection**
 Lynn Jorde, University of Utah, USA
- 10:00 **Sifting disease-causing signal from genomic noise**
 Daniel MacArthur, Massachusetts General Hospital, Boston, USA
- 10:30–11:00 **Coffee**
 Chair: Leslie Biesecker
- 11:00 **De novo diagnostics of patients with intellectual disability**
 Joris Veltman, Radboud University Nijmegen Medical Centre, Netherlands
- 11:30 **First year experience with the introduction of clinical whole exome sequencing**
 Sharon Plon, Baylor College of Medicine, USA
- 12:00 **Selected talk - Clinical diagnostic whole genome sequencing in a paediatric population; experience from our WGS genetics clinic.**
 Elizabeth Worthey, The Medical College of Wisconsin, USA
- 12:15 **Selected talk - Characterizing epistatic hotspots of human disease**
 Tallulah Andrews, MRC Functional Genomics Unit, University of Oxford, UK
- 12:30 **Sponsored technical presentation – BIOBASE**
 Jennifer Hogan, VP of Product Management, BIOBASE
- 12:30–13:45 **Lunch. Posters available**
Session 2. Ethics: dealing with data
 Chair: Sharon Plon
- 13:45 **How to avoid one thousand opportunities to do harm in genomic medicine**
 Isaac Kohane, Harvard Medical School and Children's Hospital Boston, USA
- 14:15 **Analyzing Genomes: is there a duty to disclose?**
 Amy McGuire, Baylor College of Medicine, USA
- Session 3. From discovery sequencing to targeted therapy in cancer**
- 14:45 **Interrogating of cancer genomes: towards more profile-based therapeutics**
 John Carpten, TGen, USA
- 15:15 **The implications of clonal genome evolution for cancer medicine**
 Samuel Aparicio, BC Cancer Research Centre, Canada
- 15:45–16:15 **Coffee**
- 16:15 **Selected talk - Surname leakage from personal genomes**
 Yaniv Erlich, Whitehead Institute for Biomedical Research, USA
- 16:30 **Selected talk - Multi-level genomic profiling of prostate cancers reveals a landscape spanning ageing and cancer**
 Johan Lindberg, Karolinska Institutet, Stockholm, Sweden
- 16:45 **Keynote - Genomics – Catching up to human genetics**
 Richard Gibbs, Baylor College of Medicine, USA
- 17:30–19:00 **Poster session 2 with pre-dinner drinks (even-numbered posters to be presented)**
- 19:30 **Conference dinner at MIT museum (for pre-registered delegates)**

CONFERENCE PROGRAM

DAY 3 SATURDAY 29 SEPTEMBER

- 07:45–08:55 **Coffee/breakfast.**
Session 3 continued. From discovery sequencing to targeted therapy in cancer
Chair: Peter Laird
- 09:00 **Translating cancer genomes**
Lynda Chin, MD Anderson Cancer Center, USA
- 09:30 **Genome evolution during progression to breast cancer**
Arend Sidow, Stanford University, USA
- 10:00 **Selected talk - MutaScope: a high sensitivity variant caller dedicated to high-throughput PCR amplicons sequencing**
Yost Shawn, University of California San Diego, USA
- 10:15 **Analysis of somatic retrotransposition in human cancers**
Peter Park, Harvard Medical School, USA
- 10.30 – 11.00 **Coffee**
Session 4. Epigenomics technologies and applications
Chair: Oliver Rando
- 11:00 **Exploring the Cancer Methylome**
Peter Laird, University of Southern California, USA
- 11:30 **The hunt for mammalian epialleles**
Vardhman Rakyen, Barts and The London School of Medicine and Dentistry, UK
- 12:00 **Folding principles of genomes**
Job Dekker, University of Massachusetts Medical School, USA
- 12:30 – 13:45 **Lunch. Posters available.**
- 13:45 **Ultra-high resolution mapping of protein-genome interactions using ChIP-exo**
Frank Pugh, Penn State University, USA
- 14:15 **Selected talk - Epigenetic reprogramming in the epithelial-to-mesenchymal transition**
Stephen Hoang, University of Virginia School of Medicine, USA
- 14:30 **Selected talk - Development of a computational strategy to compare repetitive element enrichment between experimental conditions from high throughput sequencing datasets**
Steven Criscione, Molecular Biology, Cell Biology, and Biochemistry, Brown University, USA
- 14:45 **Keynote - Towards a patient-based drug discovery**
Stuart Schreiber, Broad Institute of Harvard and MIT, USA
- 15.30 **Closing remarks**