



# Genome Science 2015

University of Birmingham, 7-9th September

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# WELCOME

## Dear Delegates

Our warmest welcome to the University of Birmingham for the 6<sup>th</sup> Genome Science meeting. First held in Nottingham in 2010, we have always aimed to showcase the latest developments in genomics and sequencing technology, first-class genome biology (*Is genomics biology?*) and case studies in applied and translational genomics. This meeting, our largest yet, will be no exception. Many thanks to our organisation team: Hannah Eno, Jennie Law and Catherine Wardius and our team of student volunteers. We are also grateful for the generous support of our sponsors who help keep the cost of attendance low. Please ensure you visit all the stands and find out about the products and services on offer. The speaker line-up is incredibly strong, and I know this will be a fantastic meeting. Your feedback on how we can improve in the future is always appreciated. Genome Science 2016 will be held at the University of Liverpool.

I hope you have a great meeting. Regards, Nick Loman *on behalf of the Organising Committee:*

Aziz Aboobaker, Holly Bik, Chris Ponting, Mike Quail and Mick Watson

# PROGRAMME

## Monday 7 September

1300 – 1400    **Arrival, Registration and Lunch**  
Great Hall, Aston Webb Building

### Keynote speakers

Elgar Concert Hall, Bramall Music Building

*Chair: Nick Loman, MRC Independent Research Fellow in Biomedical Informatics, Institute of Microbiology and Infection, University of Birmingham*

- 1400            Daniel MacArthur, Assistant Professor and Group Leader, Analytic and Translational Genetics Unit, Massachusetts General Hospital  
*From big data to biology: lessons from 60,000 human exomes*
- 1445            Bill Hanage, Associate Professor, Department of Epidemiology, Harvard School of Public Health  
*Is genomics biology?*

**1530            Coffee - Great Hall**

### Plenary: New Technologies

Elgar Concert Hall, Bramall Music Building

*Chair: Mike Quail, Sequencing R&D Team Leader Wellcome Trust Sanger Institute*

- 1600            **Keynote Speaker:** Mark Akeson, Professor Biomolecular Engineering, Co-Director, UCSC Biophysics Laboratory, University of California Santa Cruz  
*Nanopore sequencing for DNA and protein sensing*
- 1645            Daniel Turner, Senior Director of Applications, Oxford Nanopore Technologies  
*Oxford Nanopore Updates*
- 1715            Scott Brouillette, Senior Marketing Specialist, Illumina

## Illumina Updates

- 1730 Michael Schnall-Levin, VP of Computational Biology and Applications, 10X Genomics  
*Unlock Powerful Genomics Insights with Linked-Reads: Introducing the GemCode Platform*
- 1745 Cameron Frayling, Base4  
*Caution! May contain nudity!*

## 1815 - 1900 Posters and drinks – Great Hall

Followed by a barbecue at the Vale (accommodation location)

## Tuesday 8 September

### 0900 Coffee, Great Hall

#### Parallel Session 1: Environmental Genomics

Arts Lecture Theatre, Arts Building

*Chair: Holly Bik, Birmingham Fellow, School of Biosciences, University of Birmingham*

- 0930 Luisa Orsini, Lecturer in Biosystems and Environmental Change, University of Birmingham  
*Only time can tell: studying long-term evolutionary dynamics using biological archives*
- 1000 Douglas Yu, Professor of Ecology, University of East Anglia  
Metabarcoding and mitogenomics for endangered species conservation and management
- 1030 **Abstract-selected talk:** Martin Ayling, The Genome Analysis Centre  
*Metagenomic assembly with viral genomes*
- 1045 **Abstract-selected talk:** Bede Constantinides, University of Manchester  
*Characterising highly diverse RNA viromes in geographically distributed Venezuelan bats*

#### Parallel Session 2: Single Cell Genomics

Lecture Theatre 1, Arts Building

*Chair: Neil Hall, Professor of Functional and Comparative Genomics, University of Liverpool*

- 0930 Sarah Teichmann, Research Group Leader, European Bioinformatics Institute, European Molecular Biology Laboratory  
*Understanding cellular heterogeneity*
- 1000 David Starns (RIKEN/University of Liverpool)  
*Gotta Catch 'Em All – using single cell genomics to unravel termite nutritional symbioses*
- 1030 **Abstract-selected talk:** Harry Noyes, University of Liverpool  
*Direct discovery of bovine haplotypes by sequencing from single chromosomes*
- 10:45 **Abstract-selected talk:** Justina Zuraskiene, Wellcome Trust Centre for Human Genetics, University of Oxford  
*pcaReduce: combining k-means clustering and principal components analysis for hierarchical clustering of single cell transcriptional profiles*

### 11.00 – 11.30 Coffee – Arts Building

#### Parallel Session 3: Functional Genomics

Arts Lecture Theatre, Arts Building

*Chair: Aziz Aboobaker, Associate Professor in Genetics, University of Oxford*

1130	Amy Buck, Wellcome Trust Research Career Development Fellow, Centre for Immunity, Infection and Evolution, University of Edinburgh <i>RNA and vesicle secretion by helminths: a new mode of parasite-host communication?</i>
1200	Tatjana Sauka-Spengler, Associate Professor of Genome Biology, Radcliffe Department of Medicine, University of Oxford <i>Pioneering the chromatin for neural crest specification</i>
1230	Anura Rambukkana, Professor of Regeneration Biology and Group Leader, Centre for Regenerative Medicine, University of Edinburgh <i>Bacterial-induced epigenetic reprogramming of adult tissue cell fate</i>

#### Parallel Session 4: Bioinformatics Showcase

Arts Lecture Theatre, Arts Building

Chair: Emily Richardson, Research Fellow, MicrobesNG, Institute of Microbiology and Infection, University of Birmingham

1130	<b>Sponsored Talk:</b> Dr Tim Bonnert, Associate Director of QIAGEN Advanced Genomics <i>Sample to Insight with QIAGEN bioinformatics</i>
1150	<b>Abstract-selected talk:</b> Mohammed-Amin Madoui, Genoscope, France <i>Microbial genome assembly using synthetic error-free reads</i>
1205	<b>Abstract-selected talk:</b> Matt Loose, University of Nottingham <i>Running and Reading in Real Time: Looking at Squiggles on the Oxford Nanopore minion</i>
1220	<b>Abstract-selected talk:</b> Tom Connor, University of Cardiff <i>The Cloud Infrastructure for Microbial Bioinformatics (CLIMB)</i>
1235	<b>Abstract-selected talk:</b> Richard Leggett, TGAC <i>Flexible, multi-reference software for pre and post-alignment analysis of Nanopore sequencing data, quality and error profiles</i>

**1300 – 1400 Lunch – Great Hall**

#### Parallel Session 5: Plant and Animal Genomics

Lecture Theatre 1, Arts Building

*Chair: Mick Watson, Head of Bioinformatics, Edinburgh Genomics, The Roslin Institute, University of Edinburgh*

1400	Catherine Kidner, Professor of Plant Evolution and Development; Speciation Genetics, University of Edinburgh/Royal Botanic Gardens Edinburgh <i>Genetics and genomics of Begonia and understanding diversity</i>
1430	Paul Flicek, Senior Scientist and Team Leader, Vertebrate Genomics, European Bioinformatics Institute, European Molecular Biology Laboratory <i>Enhancers and the evolution of mammalian transcriptional regulation</i>
1500	<b>Abstract-selected talk:</b> Paul Bailey, The Genome Analysis Centre, New Online Resource Provides Millions of Induced Mutations for Gene Function Analysis in Bread Wheat
1515	<b>Abstract-selected talk:</b> Adam Hargreaves, University of Oxford, <i>Characterisation of snake venom gland transcriptomes using the Oxford Nanopore MinION portable single-molecule nanopore sequencer</i>

## Parallel Session 6: Microbial Genomics

Arts Lecture Theatre, Arts Building

**Chair:** Dr Nick Loman, MRC Independent Research Fellow in Biomedical Informatics, Institute of Microbiology and Infection, University of Birmingham

- 1400 Jukka Corander, Professor, Faculty of Mathematics and Natural Sciences, University of Helsinki  
*Being the clairvoyant: Sequence Element Enrichment (SEER) analysis for deciphering genetic basis of bacterial phenotypes*
- 1430 Josh Quick, Bioinformatician, School of Biosciences, University of Birmingham  
*Portable Nanopore sequencing for pathogen sequencing*
- 1450 Lauren Cowley, PhD student, Public Health England  
*Sequencing Ebola in the field: a tale of nanopores, mosquitoes and whatsapp*
- 1510 **Abstract-selected talk:** Tom Connor, Cardiff University  
*Using genomics to define, explore and expand our understanding of the species Shigella flexneri*
- 15:20 **Abstract-selected talk:** Andrea Gori, PhD Student, University of Exeter  
*The distribution and composition of Campylobacter jejuni plasmid pan-genome*
- 1530 Coffee – Great Hall**

## Vendor sessions

Lecture Theatre 1 Arts Building

Chair: Christiane Hertz-Fowler, Centre Manager, Centre for Genomics Research, University of Liverpool

- 1600 Dr Herbert van den Berg, KAPA Biosystems  
*Evolved enzymes for NGS library preparation*
- 1620 Lynne Apone, Ph.D , New England Biolabs  
*Novel Solutions for Challenging NGS Samples*
- 1640 Beckman Coulter
- 1650 Steve Hughes, Hamilton Robotics  
*Genomic Automation – Rigid Implementation in a Flexible World*
- 1700 Thermo Fisher
- 1710 Sudipto Das, Roche  
*Exploring the genome and epigenome of metastatic colorectal cancer using a sequence capture strategy*
- 1720 Cambridge Biosciences  
*New High Performance Methods for RNA-seq and cfDNA sequencing*

## Conference ‘Street Food’ dinner, Alfie Birds, Custard Factory, Digbeth

Transport provided from Great Hall, coaches at 6pm from outside Great Hall

# Wednesday 9 September

0900 Coffee, Great Hall

## Parallel Session 7: Clinical Genomics

Arts Lecture Theatre, Arts Building

**Chair:** Andrew Beggs, Clinical Lecturer in General Surgery, School of Cancer Sciences, University of Birmingham

- 0930 Sarah Ennis, Professor of Genomics, School of Medicine, University of Southampton  
*Paediatric exomes: diagnostic and data dilemmas*
- 1000 Pippa Thomson, Lecturer, Centre for Genomic and Experimental Medicine, School of Molecular, Genetic and Population Health Sciences, University of Edinburgh  
*Using high-throughput sequencing to understand the genetic complexity of psychiatric illness*
- 1030 Anthony Rogers, Congenica  
*Why we care about rare diseases*
- 1045 **Abstract-selected talk:** Saskia Sanderson, Department of Genetics and Genomic Sciences, Icahn School of Medicine, New York, NY, *Should everyone have their genomes sequenced? Insights from interviews with personal genomics research participants*
- 1100 Coffee – Great Hall

## Parallel Session 8: Computational Genomics

Lecture Theatre 1, Arts Building

**Chair:** Chris Ponting, Professor of Genomics at the University of Oxford and an Associate Faculty member at the Wellcome Trust Sanger Institute

- 0930 Martin Taylor, Group Leader, Biomedical Systems Analysis, Institute of Genetics and Molecular Medicine, MRC Human Genetics Unit, University of Edinburgh Western General Hospital  
*The frequent evolutionary birth and death of functional promoters*
- 1000 Gosia Trynka, Group Leader, Wellcome Trust Sanger Institute, Immune Genomics  
*Using epigenetics for biological interpretation of complex trait associations*
- 1030 **Abstract-selected talk:** Nicholas J Harding, Wellcome Trust Centre for Human Genetics, University of Oxford  
*Challenges of fine and broad scale diversity in the genome of the malaria vector Anopheles gambiae*
- 10:45 **Abstract-selected talk:** Mick Watson, Head of Bioinformatics, Edinburgh Genomics  
*Is this the end of RNA-Seq alignment? Benchmarking RNA-Seq quantification tools*

## Plenary session

Elgar Concert Hall, Bramall Music Building

**Chair:** Nick Loman, MRC Independent Research Fellow in Biomedical Informatics, Institute of Microbiology and Infection, University of Birmingham

- 1130 Inês Barroso Head of Human Genetics at the Wellcome Trust Sanger Institute and leader of the Metabolic Disease Group  
*Genomics and Metabolic Disease – what have we learned so far?*
- 1215 Matthew Cobb, University of Manchester  
*Life's greatest secret: the story of the race to crack the genetic code*
- 1300 Conference close

**Dr Matthew Cobb will be signing copies of his new book in the coffee area of the Bramall Building after his talk.**

# Posters

1. Laura Oikkonen, University of Oxford  
*Making the most of RNA-seq: Pre-processing sequencing data for reliable SNP variant detection*
2. Stefan Bagby, University of Bath  
*Use of MinION data to assemble the 67% GC genome sequence of *Bordetella pertussis**
3. Dagmar Frisch, University of Oklahoma  
*Paleogenetic records of *Daphnia pulicaria* in two North American lakes reveal the impact of cultural eutrophication*
4. M.W.Wojewodzic, University of Birmingham  
*Seeking for Transgenerational Effects of Diet Quality in Clones –Ecophysiological and RNA-seq approaches.*
5. Tamsin M. O. Majerus, University of Nottingham  
*Investigation of the genetic control of colour-pattern in the 2- and 10-spot ladybirds*
6. Lisa Crossman, University of East Anglia  
*Marine Streptomyces genomes from Illumina and Nanopore sequences*
7. Gloria Despacio-Reyes, Wellcome Trust Sanger Institute  
*Using new transcriptomic data to improve the annotation of long non-coding RNA genes*
8. Bhavana Harsha, Wellcome Trust Sanger Institute  
*Comparative genomics of *Onchocerca volvulus**
9. Katherine Fawcett, University of Oxford  
*De novo point mutations in patients diagnosed with ataxic cerebral palsy*
10. Deepa Manthravadi, Wellcome Trust Sanger Institute  
*The HAVANA Manual Annotation of Vertebrate Genomes: creating comprehensive gene sets for human and mouse*
11. Georgia Kapatai , Public Health England  
*Molecular pneumococcal capsular typing using whole genome sequencing: moving the *Streptococcus pneumoniae* reference service into the genomic era.*
12. Michael Clark, University of Oxford  
*Quantitative gene expression profiling with targeted RNA sequencing*
13. Matthew Gemmell, University of Aberdeen  
*Comparison of 16S rRNA v1-2 and v3-4 regions for bacterial community analysis in stool samples using Illumina MiSeq V3 chemistry*
14. Reema Singh, University of Dundee  
*Refined Transcriptome Annotation via de novo Assembly in Two Social Amoeba Species*
15. Adam Peltan, New England Biolabs  
*Repair of challenging FFPE DNA improves library success rate and sequencing quality*
16. Chris Lounds, New England Biolabs  
*Hemoglobin Depletion Increases Sensitivity of Next Generation Sequencing-based Profiling*
17. Lynne Apone, New England Biolabs  
*High Quality Library Construction and Reliable Quantitation with NEBNext Reagents*
18. Fiona Stewart, New England Biolabs  
*NGS Library Preparation Method for Transcriptome Profiling With Enhanced Sensitivity of Transcript Detection*
19. Ian Sudbery, University of Sheffield  
*The trouble with UMIs*
20. Avraam Tapinos , University of Manchester  
*Alignment by numbers: Sequence assembly using compressed numerical representations*
21. Nicole Gruenheit, University of Manchester  
*REMI-seq – a step change in *Dictyostelium* functional genomics*
22. Dave Baker, The Genome Analysis Centre  
*High Throughput Development of Pacbio 20kb Template Preparation*
23. Judith E. Risso, University of Edinburgh  
*Catching shufflons in the act: Using nanopore reads to study *Bacteroides fragilis**
24. Oliver Deusched, Waltham Petcare  
*The adolescent feline faecal microbiome: a longitudinal study into the effects of age,*
25. Neil Pearson, The Genome Analysis Centre  
*3D printing for rapid laboratory prototyping*
26. Darren Heavens, University of Edinburgh  
*A method to simultaneously construct up to 12 different sized Illumina Nextera long mate pair libraries with reduced DNA, time and costs*
27. Teri Evans, University of Nottingham  
*The genome of the salamander *Ambystoma mexicanum**
28. Bert Overduin, University of Edinburgh  
*Bioinformatics Training for Genomics – One year on*



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