2022 Genetics Society Meeting

Functional Regulatory Genomics & Disease

11 – 13 April 2022, Surgeons' Hall, Edinburgh, UK and Online

Meeting Programme

the geneticssociety

WELCOME

It is my pleasure, on behalf of the organising committee, to welcome you all to Edinburgh and our first conference on Functional Regulatory Genomics and Disease.

We will be gathered in the famous Surgeons' Hall, established in 1699, that acts as the headquarters of the Royal College of Surgeons of Edinburgh (itself founded in 1505). Surgeons' Hall is an historic grade-A listed building and will provide an amazing venue for our conference. I hope that some of the less squeamish amongst you can take the opportunity to explore the world-famous collections held in the Hall.

The focus of the conference will be on the role of the enigmatic non-coding regulatory genome in human health and disease. Our first session, chaired by Prof Andrew McIntosh (Edinburgh University), will explore the evidence, from human GWAS studies, that the non-coding regulatory genome plays a role in health and disease.

The second session, chaired by Prof Boris Lenhard (Imperial College, London), will examine how the expanding data sets produced by next generation sequencing based technologies is permitting a greater understanding of the extent and function of the non-coding genome in health and how it can go wrong in disease.

The third session, to be chaired by Prof Wendy Bickmore (MRC Human Genetics Unit, Edinburgh), will focus on the functional biology of known components of the regulatory genome, that include enhancers, promoters and insulators, and the molecular interactions that they require to fulfil their functions within living systems.

The fourth and final session will be chaired by Dr Michelle Holland (King's College, London) and will examine the influence of the environment on the function of the non-coding genome through processes that include DNA and histone methylation, and how this may impact on health and disease susceptibility.

However, the main purpose of the meeting is to get people from different strands of the field mingling, talking and, yes even arguing, about the best ways to develop our understanding of the roles of gene regulation in human health and disease. By doing so, it is hoped that new insights, perceptions, and ideas may be swapped and gleaned, and new fruitful collaborations formed.

We would like to thank the Genetics Society and the Royal Society of Biology for supporting the conference and Kay Boulton, Harriet McAra and Karen Patel for organising the conference at such short notice in Edinburgh when all hope had nearly disappeared.

Kindest regards,

Dr Alasdair MacKenzie, University of Aberdeen, on behalf of the conference organising committee.

PROGRAMME

Monday 11 April 2022

12:00	REGISTRATION AND LUNCH, KING KHALID RECEPTION
13:00	WELCOME AND INTRODUCTION TO THE CONFERENCE DR ALASDAIR MACKENZIE, UNIVERSITY OF ABERDEEN

SESSION 1: THE NON-CODING GENOME IN HEALTH AND DISEASE

Chair: Professor Andrew MacIntosh, University of Edinburgh

13:05	**Professor Andrew McIntosh, University of Edinburgh** Introduction to the session
13:10	**Professor Andrew McIntosh, University of Edinburgh** The genetic architecture of depression
13:35	**Professor Ruth Loos, Icahn School of Medicine at Mount Sinai** The genetics of obesity – from genes to biology… to clinical practise
14:00	**Dr Erola Pairo-Castineira, Roslin Institute, Edinburgh** Genetic mechanisms of critical illness in Covid-19
14:25	REFRESHMENT BREAK
14:45	**Professor Marcelo Nobrega, University of Chicago** Functionally dissecting the genetic landscape of complex human traits
15:10	**Professor Chris Ponting, University of Edinburgh** Pinpointing casual variants for human complex traits and diseases
15:35	**Dr Eilis Hannon, University of Exeter** Determining cell-specificity of epigenetic variation associated with brain disorders
16:00	**Maria Lillina Vignola, King's College London** A multilayer experimental strategy to localise transcriptional regulatory elements involved in the pathogenesis of human fetal growth restriction
16:15	**Liane Fernandes, Queen Mary University of London** A satellite DNA array barcodes chromosome 7 and regulates totipotency via ZFP819
16:30	**Poster pitches session** Introduction by Dr Laura Lettice, University of Edinburgh
	Grace Alston, Wellcome Centre for Cell Biology Edinburgh Decoding PAX6 Enhancer Grammar Using Regressive Evolution of the Eye
	Dr Fanny Boulet, Blizard Institute BRD4 mutation promotes neurogenesis and inhibits oligodendrocytes and glial cells
	James Cain, King's College London Utilising an imprinted locus to study the interplay between intragenic transcription and alternative polyadenylation
	Michael Claxton, University of Liverpool Variability and mosaicism in allelic expression of imprinted genes in single neural cells - comparison to whole brain tissue

Alexander Fröhlich, University of Liverpool
Characterisation of the Function of a SINE-VNTR-Alu Retrotransposon to Modulate Isoform Expression at the MAPT Locus

Katy Graham, University of Edinburgh
Transcriptional dynamics of the Sonic hedgehog gene

Victoria Lindsay, Royal Veterinary College
The role of the regulatory genome in complex metabolic diseases: the case of equine exertional rhabdomyolysis

Dr Debosree Pal, Blizard Institute
Role of MSL-mediated H4K16ac in mammalian genome regulation

Adrian Rodriguez, Queen Mary University of London
Epigenetic mechanisms in atrial fibrillation: the role of developmental regulators

Kirsty Uttley, University of Edinburgh
Decoding the functional properties of overlapping PAX6 retinal enhancers in vivo

17:00 DRINKS RECEPTION AND POSTER VIEWING
PLAYFAIR HALL, PLAYFAIR BUILDING

Tuesday 12 April 2022

08:30 REGISTRATION, KING KHALID RECEPTION

SESSION 2: "BIG DATA" APPROACHES IN FUNCTIONAL GENOMICS

Chair: Professor Boris Lenhard, University College London

09:00 **Professor Boris Lenhard, University College London**
Introduction to the session

09:05 **Professor Tatjana Sauka-Spengler, University of Oxford**
Deciphering gene regulatory networks in development and disease

09:30 **Professor François Spitz, Institut Pasteur**
Genomics and epigenomics of animal development

09:55 **Professor Jim Hughes, University of Oxford**
Decoding the non-coding genome in health and disease

10:20 REFRESHMENT BREAK

10:40 **Professor Stefan Hoppler, University of Aberdeen**
WNT signalling in early vertebrate embryogenesis and human heart muscle differentiation

11:05 **Professor Boris Lenhard, University College London**
Responsiveness of promoters to enhancers in long-range developmental regulation

11:30 **Dr Renée van Amerongen, University of Amsterdam**
Tissue-specific WNT signaling: Dissecting the mechanisms that control WNT gene expression in the breast

11:55 **Dr Devin Bendixsen, University of Edinburgh**
Identifying driver variants within the mutational landscape of high grade serous ovarian cancer

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Tuesday 12 April 2022

12:10	**Dr Nicolae Radu Zabet, Queen Mary University of London** Using Explainable Artificial Intelligence to decipher the epigenetic code of enhancers
12:30	LUNCH BREAK POSTER SESSION

SESSION 3: THE BIOLOGY OF GENE REGULATION

Chair: Professor Wendy Bickmore, MRC Human Genetics Unit, Edinburgh

13:30	**Professor Wendy Bickmore, MRC Human Genetics Unit, Edinburgh** Introduction to the session
13:35	**Professor Robert Hill, University of Edinburgh** Mutations in a long distance regulator effects congenital defects
14:00	**Dr Alasdair MacKenzie, University of Aberdeen** Exploring the regulatory genetics of fat and alcohol intake and anxiety
14:25	**Dr Emma K Farley, University of California San Diego** Affinity optimizing mutations disrupt development
14:50	**Professor Marc S Halfon, University at Buffalo** Convergence and divergence of enhancer sequences
15:15	REFRESHMENT BREAK
15:35	**Professor Axel Visel, Lawrence Berkeley National Laboratory** Distant acting enhancers in development, disease and evolution
16:00	**Professor Wendy Bickmore, MRC Human Genetics Unit, Edinburgh** Enhancer-promoter communication: is close enough, enough?
16:25	**Professor Douglas Higgs, University of Oxford** Gene regulation during haematopoiesis
16:50	**Dr Hannah Long, University of Edinburgh** Regulatory and topological perturbation at a human disease locus
17:05	**Dr Ariella Weinberg-Shukron, University of Cambridge** Balanced Gene Dosage Control rather than Parental Origin underpins Genomic Imprinting
19:00	CONFERENCE DINNER INTERCONTINENTAL EDINBURGH THE GEORGE HOTEL

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Wednesday 13 April 2022

08:30 REGISTRATION, KING KHALID RECEPTION

SESSION 4: EPIGENETICS AND GENE REGULATION

Chair: Dr Michelle Holland, King's College London

09:00 **Dr Michelle Holland, King's College London**
Introduction to the session

09:05 **Dr Chris Murgatroyd, Manchester Metropolitan University**
Epigenetics of early life stress

09:30 **Dr Kathy Evans, University of Edinburgh**
Characterising DNA methylation signatures of dementia risk and of alcohol consumption

09:55 **Professor Gert Jan Veenstra, University of Nijmegen**
The role of Polycomb in gene regulation and exit of pluripotency

10:20 REFRESHMENT BREAK

10:35 **Professor Rebecca Oakey, King's College London**
The activity of intragenic CpG islands influences alternative splicing and polyadenylation genome-wide generating tissue-specific transcriptomes

11:00 **Professor John Quinn, University of Liverpool**
Endogenous retrotransposon polymorphisms; their influence on gene expression and disease

11:25 **Dr Michelle Holland, King's College London**
Non-coding variation associated with BMI in humans

11:50 **Dr Poppy Gould, Queen Mary University of London**
An ancient KRAB-zinc finger protein, ZFP37, targets active endogenous retroviruses and regulates neural development

12:05 **Dr Stephanie Frost, Queen Mary University of London**
Cardiovascular trait-associated variants and their interplay with epigenomic enhancer conservation

12:20 **Dr Alasdair MacKenzie, University of Aberdeen**
Final comments

12:30 LUNCH BREAK

A visit to the Surgeons' Hall Museum is highly recommended, entry £8.50

19:00 **Public engagement event**
Deacon's Suite
Dr Alasdair MacKenzie, University of Aberdeen
Genomic "Dark Matter" and its role in human health and disease

www.genetics.org.uk