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Edinburgh-St Andrews Consortium for Molecular Pathology, Informatics and Genome Sciences

International Molecular Pathology Symposium

Tuesday 31st May 2016
09.00 - 18.00
IGMM Lecture Theatre
University of Edinburgh

KEYNOTE SPEAKERS

Professor Pancras Hogendoorn
University of Leiden

Professor Joakim Lundeberg
SciLifeLab, Stockholm

Dr David Bentley
Illumina, Inc

Professor Sue Hill
Chief Scientific Officer England

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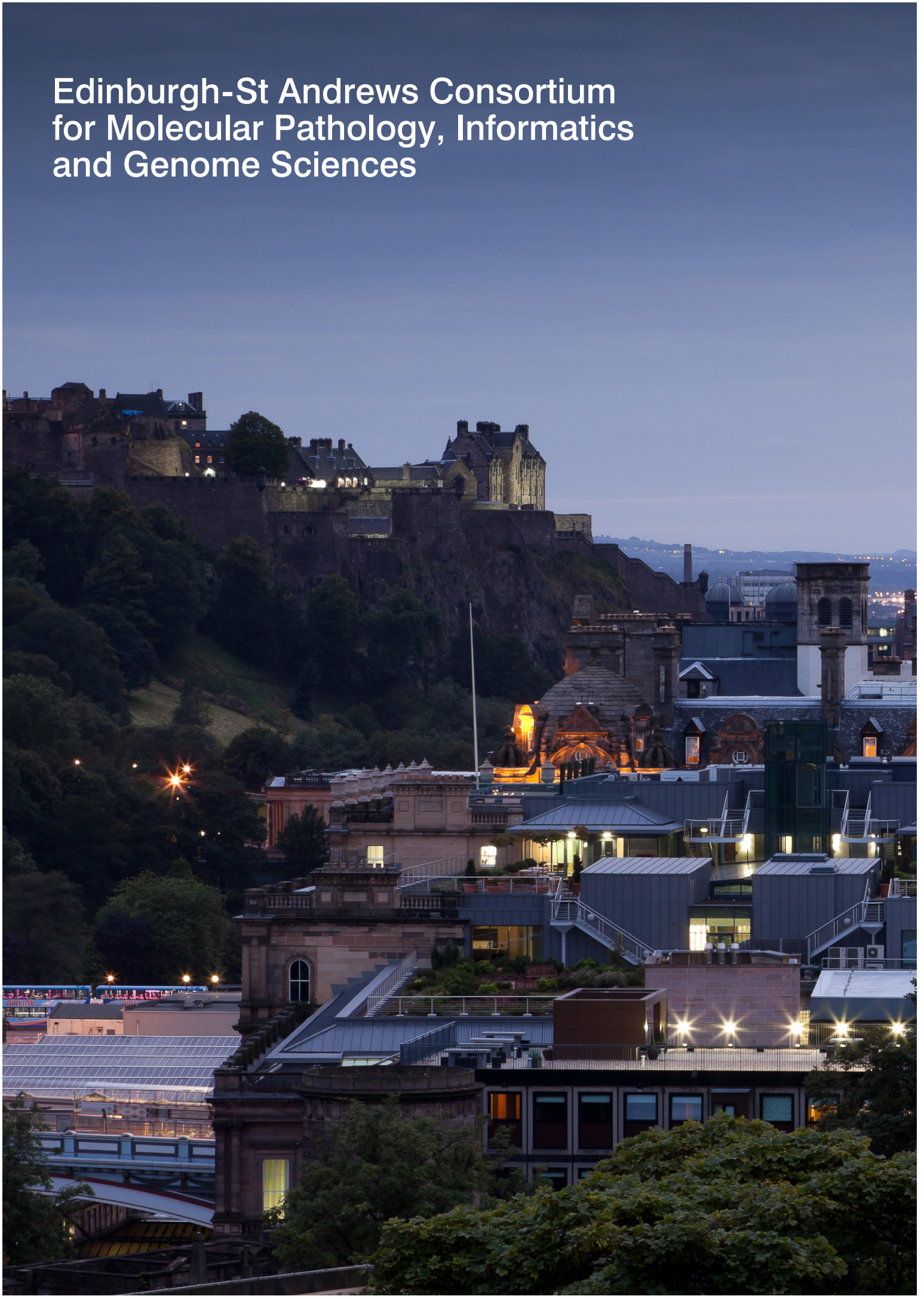

International Molecular Pathology Symposium
Programme

PROGRAMME

Tuesday 31st May 2016 IGMM Lecture Theatre, The University of Edinburgh

09.00 **Coffee and Registration**

09.30 **Professor Sir John Savill**
Welcome and Introductory remarks
VP, Head of the College of Medicine & Veterinary Medicine, University of Edinburgh, Chief Executive, MRC

09.45 **Professor Sue Hill**
Keynote: Integrating genomics - the future of medicine
Chief Scientific Officer, NHS England

Genomics, Informatics and Pathology (1) Chair: Professor Mary Porteous
University of Edinburgh

10.15 **Professor Simon Herrington**
University of Edinburgh, The Edinburgh-St Andrews Molecular Pathology Node

10.25 **Dr Karin Oien**
University of Glasgow, Glasgow Molecular Pathology Node and CMPath

10.35 **Professor Tim Aitman**
University of Edinburgh,
Scottish Genomes Partnership

10.50 Tea/Coffee

Genomics, Informatics and Pathology (2) Chair: Professor Mark Arends
University of Edinburgh

11.20 **Professor Pancras Hogendoorn**
University of Leiden
Keynote: Molecular Pathology of Sarcomas: From genes to understanding disease

11.50 **Professor Malcolm Dunlop**
University of Edinburgh, Molecular pathology of colorectal cancer risk

12.10 **Dr Clare Turnbull**
Genomics England Ltd, 100,000 Genomes Cancer Programme: opportunities around molecular pathology

12.30 **Professor Colin Semple**
University of Edinburgh,The blind watch-breaker: evolution at regulatory sites in cancer

12.50 Lunch and Molecular Pathology MMedSci Student Posters

Pathology, Imaging and other 'omics
Chair: Professor David Crossman
University of St Andrews

13.50 **Dr Kev Dhaliwal**
University of Edinburgh, Optical Molecular Sensing and Imaging of Human Disease In Situ

14.10 **Professor Jürgen Haas**
University of Edinburgh, Systematic analysis of host factors affecting virus infections

PROGRAMME

Tuesday 31st May 2016 IGMM Lecture Theatre, The University of Edinburgh

14.30 **Professor Angus Lamond**
University of Dundee, Multidimensional Proteomic Analysis of Disease Mechanisms

14.50 **Professor Joakim Lundeberg**
SciLifeLab, Stockholm
Keynote: Spatially resolved gene expression patterns in tissue sections

15.20 Tea/Coffee

Diagnostics, Therapeutics and industry collaboration
Chair: Dr Marc Muskavitch, Biogen

15.50 **Professor Andrew Biankin**
University of Glasgow, Precision Oncology and the Scottish Genomes Partnership

16.10 **Dr Sandi Deans**
UK NEQAS, Ensuring high quality Molecular Pathology testing

16.30 **Dr David Bentley**
Illumina, Inc.
Keynote: Genomes for Medicine

17.00 **Professor Simon Herrington & Professor Tim Aitman**
Closing comments

17.15 Drinks Reception

Edinburgh-St Andrews Consortium for Molecular Pathology, Informatics and Genome Sciences

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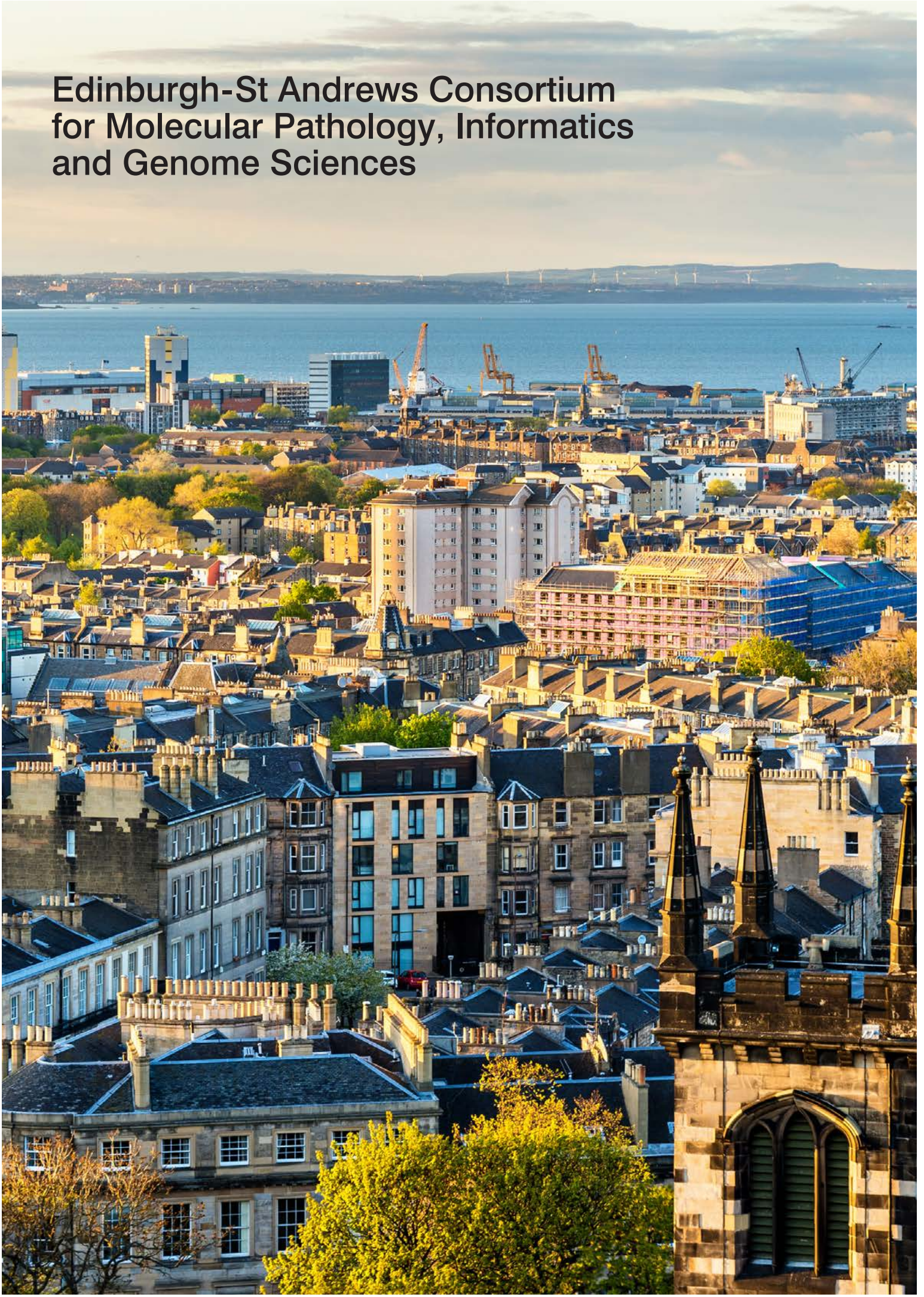

International Molecular Pathology Symposium
Speaker Profiles

Professor Tim Aitman

Professor Tim Aitman is the Director of the Centre for Genomic and Experimental Medicine within the MRC Institute of Genetics and Molecular Medicine. He is Professor of Molecular Pathology and Genetics at the University of Edinburgh and Director of the Edinburgh-St Andrews Consortium for Molecular Pathology, Informatics and Genome Sciences, one of only six new MRC-EPSRC Molecular Pathology Nodes in the UK. Professor Aitman is the co-Director and Edinburgh PI of the Scottish Genomes Partnership, Clinical Director of the HighSeqX facility in Edinburgh Genomics and also a Consultant Physician in NHS Lothian.

A graduate of Birmingham Medical School and Kings College London he obtained his DPhil at Wolfson College, Oxford University. Before joining the University of Edinburgh in 2014, he was Group Head and Section Chair at the MRC Clinical Sciences Centre, Hammersmith Hospital, Honorary Consultant Physician at Hammersmith Hospitals NHS Trust and Professor of Clinical & Molecular Genetics in the Imperial College London Faculty of Medicine (where he continues as Visiting Professor). Prof Aitman is a Fellow of the Royal College of Physicians, Academy of Medical Sciences and Society of Biology. He is also a Trustee of the Public Health Genomics (PHG) Foundation and was the Specialist Adviser for the House of Lords Science and Technology Committee's Inquiry into Genomic Medicine.

Professor Aitman has authored over a hundred scientific papers, many highly cited, and has been invited to give over 150 plenary and state-of-the-art lectures at major national and international conferences. He co-ordinated multiple scientific projects and research consortia with career grant support of over £30 million and has been a mentor of many successful graduate students and postdoctoral scientists.

Professor Mark Arends

Mark Arends trained in Medicine (MBChB with Honours) and Pathology (BSc with Honours and PhD) at the University of Edinburgh, becoming a Senior Lecturer in 1995 in the Edinburgh University Department of Pathology and Honorary Consultant at the Royal Infirmary of Edinburgh (MRCPath & FRCPath). He moved to the University of Cambridge (MA) in January 1999, where he was a University Reader in Histopathology and an Honorary Consultant at Addenbrooke's Hospital, Cambridge. There he was a specialist gastrointestinal and gynaecological pathologist, was lead pathologist in colorectal pathology, gynaecological pathology and bowel cancer screening pathology for Cambridge and East of England. In July 2013 he moved to University of Edinburgh as Professor of Pathology, Head of the Division of Pathology and Co-director of the Centre for Comparative Pathology. His research includes the genomic, genetic and epigenetic mechanisms of colorectal cancer and gynaecological cancer development and progression, including inherited susceptibility to colorectal and endometrial carcinogenesis and in vivo models of intestinal tumour formation.

Professor David Bentley

David Bentley DPhil FMedSci is Vice President and Chief Scientist at Illumina Inc.

David graduated with an M.A. in Natural Sciences from Cambridge and a D.Phil from Oxford. During his career he has been a Senior Lecturer at London University; and later the Head of Human Genetics and a founder member of the Board of Management at the Sanger Centre. David has played a leading role in the Human Genome Project and related international consortia to characterise human sequence variation, including The SNP Consortium and the HapMap Project. His long-term interest is the study of human sequence variation and its impact on human health and disease. His current research is focussed on fast, accurate sequencing of human genomes for adoption and benefit in healthcare.

Professor Andrew Biankin

Professor Andrew Biankin is a Clinician Scientist, the Director of the Wolfson Wohl Cancer Research Centre and Regius Chair of Surgery at the University of Glasgow. His primary scientific focus is on the molecular pathology of pancreatic cancer, the development of novel therapeutic strategies based on molecular phenotyping and the delineation and implementation of biomarkers that facilitate clinical decision-making. He contributes to the International Cancer Genome Consortium and the TCGA through characterising the molecular pathology of pancreatic cancer, and is extending this knowledge to a personalized model of cancer care, where molecular characteristics guide treatment decisions. He Chairs the Cancer Research UK National Pancreatic Cancer Network and is a member of NCRI Clinical Study Groups for Upper Gastrointestinal and Hepato-Pancreato-Biliary cancers, working with academia and industry to advance precision medicine for cancer.

He has authored over 100 articles in major peer reviewed journals including seminal works on pancreatic cancer, precision medicine and the clinical management of pancreatic disease and sits on several international expert panels for pancreatic cancer. He is regarded as a key opinion leader and advises government agencies and international cancer organisations. He works closely with industry through appointments to advisory boards and is principal investigator on international clinical trials. He co-founded Cure Forward Corporation, a patient-facing personalized medicine company where he is the Chief Scientific and Medical Advisor.

Professor David Crossman

David Crossman is Dean of the Faculty of Medicine and Head of the School of Medicine at the University of St Andrews. He is an honorary consultant cardiologist at NHS Fife where he has weekly clinical commitments. He moved to Scotland just under two years ago having run the medical school at UEA. He is Chair of the MRC/NIHR EME board (Translational medicine). He is interested in the pathological basis of myocardial infarction.

Dr Sandi Deans

Dr Sandi Deans is a Consultant Clinical Scientist and the Director of UK National External Quality Assessment Service (UK NEQAS) for Molecular Genetics which is based in the Department of Laboratory Medicine, Royal Infirmary of Edinburgh. The UK NEQAS scheme delivers assessment of molecular genetic testing, molecular pathology testing, newborn screening, prenatal diagnosis, preimplantation genetic diagnosis and is involved in the new technical next generation sequencing scheme.

Sandi is a Senior Honorary Lecturer in the Medical School, University of St. Andrews and is a collaborator in the Universities of Edinburgh and St Andrews Molecular Pathology Node including the joint supervisor of one of the 2016 Masters students.

She sits on many committees including the European Molecular Quality Network Board, UK NEQAS Steering Committees for Clinical Cytogenetics, Cellular Pathology Techniques and Molecular Genetics, and has recently been appointed as Board member and Treasurer of the International Quality Network for Pathology (IQN Path). She is a past UK NEQAS Executive Director, Association of Clinical Genetic Science Executive Committee member and has been involved in the selection panels for MCADD extended mutation screening laboratories and the Cancer Research UK Stratified Medicine Technical Hub laboratories.

Sandi is currently working as part of the NHS England 100,000 Genomes project implementaion unit and collaborates closely with Genomics Enlgand to deliver high quality sequencing for both the rare diseases and cancer programmes.

Dr Kev Dhaliwal

Kev Dhaliwal, MBChB (Hons), BSc (Hons), PhD, FRCP is a Senior Clinical Lecturer in Pulmonary Molecular Imaging at the MRC Centre for Inflammation Research in the Queen's Medical Research Institute (QMRI) and Consultant Physician in Respiratory Medicine. After graduating from Edinburgh Medical School, he completed general professional training in London before returning to Edinburgh to undertake a MRC Clinical Training Fellowship. He combined clinical and academic training as a clinical lecturer and developed a research theme in pulmonary optical molecular imaging. Kev is chief investigator on 5 first-in-man trials ranging from the intensive care unit to lung cancer evaluating optical imaging approaches. He leads the interdisciplinary hub of scientists in the QMRI as part of the Proteus Project (www.proteus.ac.uk), the UK's largest healthcare bio-photonics project which is an Engineering Physical Sciences Research Council Interdisciplinary Research Collaboration developing next-generation translational technologies for in situ pulmonary molecular imaging and sensing. He is the Medical School lead for the OPTIMA EPSRC and MRC Centre for Doctoral Training in Optical Medical Imaging (www.optima-cdt.ac.uk) which embeds entrepreneurship within the training programme. In 2014, he co-founded Edinburgh Molecular Imaging (EMI) (www.edinimage.com) which is developing optical molecular imaging approaches for colorectal cancer detection and intraoperative surgical guidance. As a clinical founder director for EMI, he is establishing global clinical trials of optical imaging in the US, Europe and Far East and developing networks to expedite clinical adoption.

Professor Malcolm Graham Dunlop

My research interests are themed around the ultimate aim of reducing of morbidity and mortality from large bowel cancer through early detection and prevention. I hold substantial current grant funding including Cancer Research UK Programmatic Funding and MRC grants. My primary research focus is in elucidating the genetic basis of large bowel cancer and applying genetic information to prevent the disease as well as to understand disease causation. To this end my group are investigating the status and the control of the transcriptional architecture of the large bowel epithelium and how that defines colorectal cancer risk. I have successfully supervised or co-supervised over 25 postgraduate degrees. I currently sit on various expert review and advisory committees, including the Experimental Medicine Expert Review Panel for Cancer Research UK, MRC Non-Clinical Fellowships Committee, Research Careers Committee of the Academy of Medical Sciences, IBD Specialist Advisory Board of the Association of Coloproctcology of GB&I, the Governance Board of the Edinburgh Cancer Research Centre, the Scientific Advisory Committee of the Melville Trust for the Care and Cure of Cancer. I previously sat on the MRC Molecular and Cellular Medicine Board and many Grants/Fellowships Committees, including Cancer Research UK and the Scottish Government, previously Chairing the Scottish Government Cancer Research Portfolio Steering Committee. I have participated in review panels for many of the UK major research groupings/institutes. I currently review grants for MRC, CRUK, WCRF (previously AICR), Scottish Government, Dutch Cancer Society. I also review manuscripts for many high impact journals including Nature, Nature Genetics, Nature Medicine, Nature Communications, New England Journal of Medicine and Lancet.

Professor Jürgen Haas

Jürgen Haas is the Chair of Viral Genomics and current Head of the Division of Infection and Pathway Medicine at the University of Edinburgh. He studied medicine and biology in Tubingen and Munich and then performed a postdoctoral training in Brian Seed's laboratory at the Massachusetts General Hospital in Boston. After his return to Germany he finished his medical specialist training in Microbiology and Virology in Heidelberg and Munich and became associate professor. In 2006 he was recruited to Edinburgh. His current work focuses on the identification of pathogen and host factors determining pathogenicity using large-scale genomic screens, and on genetic determinants of virus infection. He is also working as an honorary Consultant Virologist at the Royal Infirmary Edinburgh.

Professor Simon Herrington

Simon Herrington graduated in biochemistry with honours from the University of Cambridge in 1982 and medicine with honours from the University of London in 1985. He trained in Internal Medicine (MRCP 1988) and then became a CRC clinical research fellow in the Nuffield Department of Pathology, University of Oxford, receiving his DPhil in 1991. He was appointed clinical lecturer in pathology in Oxford where he completed his training in clinical cellular pathology, gaining the MRCPath in 1994. He was appointed clinical senior lecturer and consultant in pathology at the University of Liverpool in 1995 and was promoted to a Personal Chair in 1999. In 2003, he was appointed to the Chair of Pathology at the University of St Andrews, moving to the Chair of Pathology at the University of Dundee in 2010 and the Chair of Molecular Cancer Pathology at the University of Edinburgh in 2015. He also works as a consultant pathologist, specialising in gynaecological pathology, was co-editor of the 4th Edition of the WHO Classification of Tumours of Female Reproductive Organs, and edited the 15th Edition of Muir's Textbook of Pathology. He sits on the Board of Worldwide Cancer Research and is a past President of the International Society of Gynecological Pathologists. He is Editor in Chief of the Journal of Pathology and the Journal of Pathology: Clinical Research.

Professor Sue Hill OBE

Professor Sue Hill OBE PhD DSc CBiol FSB Hon FRCP Hon FRCPath is the Chief Scientific Officer for England and the head of profession for the 50,000 healthcare science workforce in the NHS and associated bodies – embracing more than 50 separate scientific specialisms. She is a respiratory scientist by background with an international academic and clinical research reputation.

Professor Hill has a broad portfolio of policy responsibilities across NHS England and the wider NHS and provides professional leadership and expert clinical advice across the whole health and care system. In particular, Sue is the Senior Responsible Officer for Genomics in NHS England has established NHS Genomic Medicine Centres and is now leading the NHS England Personalised Medicine strategy.

A significant part of her role involves working across government, with the Department of Health, with the NHS, Public Health and Health Education England and other external stakeholders to inform policy, influence legislation, deliver strategic change and to introduce new and innovative ways of working.

Professor Pancras Hogendoorn

Dr. Hogendoorn is member of the board of LUMC, Dean and Professor of Pathology at the Leiden University Medical Center. He completed his Medical and PhD studies at Leiden University followed by his residency in anatomic pathology. He completed fellowships in oncological pathology at the Dutch Cancer Institute, Amsterdam and Memorial Sloan Kettering Cancer Center, New York. For the last 18 years, his scientific research has focused on the pathogenesis, morphology and molecular pathology of bone - and soft tissue tumours.

He has had or has a number of national and international scientific and administrative positions, including chairmanship of the Netherlands Committee on Bone tumours, the Pathology and Biology subcommittee of the EORTC Soft tissue and Bone Sarcoma Group, European Osteosarcoma Intergroup, EuroEwing Consortium, and membership of the Board of the Connective Tissue Oncology Society and European Musculoskeletal Oncology Society and he served as overall Editor of the 2013 WHO edition of classification of bone tumors.

He received a membership of honour in recognition of his repeated courses on pathology of bone tumours from the Belgium Society of Pathology, the European Oswald Vander Vekenprijs (2008), The Nina Axelrad Award, The Jeremy Jass Price as well as the Corrine Farrel Award (International Skeletal Society) twice, as well as prices for postgraduate teaching. He holds a chair as visiting professor in sarcoma pathology at the University of Oxford.

Professor Angus Lamond

Professor Angus Lamond FRS FRSE FMedSci is Professor of Biochemistry in the School of Life Sciences, Centre for Gene Regulation and Expression at the University of Dundee in Scotland. Before moving to Dundee in 1995 Angus was a group leader at the European Molecular Biology Laboratory in Heidelberg, where he started using mass spectrometry based proteomics techniques. Angus' group study gene expression and the functional organization of mammalian cell nuclei, using a strategy that combines quantitative mass spectrometry (MS) based proteomics and live cell fluorescence imaging (see www.LamondLab.com). They have studied the nucleolar proteome and the regulated assembly of nucleoli in human cells. The Lamond group have developed proteomic methods for studying the dynamics of protein localization, turnover and protein-protein interactions. They have created a software project – Peptracker – aimed at the efficient integration and analysis of multiple large proteomics datasets. This is used to identify and characterize biological regulatory and disease mechanisms in both human cells and model organisms.

Professor Joakim Lundeberg

Professor Joakim Lundeberg heads the division of Gene Technology, KTH Royal Institute of Technology. His research group is since May 2010 located at the Science for Life Laboratory (SciLifeLab), a national center for molecular biosciences with focus on health and environmental research. The center combines frontline technical expertise with advanced knowledge of translational medicine and molecular bioscience. JL also heads the National Genomics Infrastructure (NGI), together with Profs Syvänen and Gyllensten, that provides Swedish researchers access to broad range of state of the art instrumentation for massively parallel sequencing and genotyping with a staff of ~70 FTEs.

Dr Marc Muskavitch

Marc Muskavitch has been involved in research that has ranged over biochemistry, genetics, cell and developmental biology, neuroscience, infectious disease, genomics and epigenetics, over the course of nearly 30 years of academic research and administration, and graduate, undergraduate and postdoctoral teaching and training, and since his move to Biogen.

After earning a B.S. in biochemistry at University of Wisconsin-Madison and a Ph.D. in biochemistry from Stanford University, he pursued postdoctoral research in genetics at Harvard University. In 1984, he accepted a tenure-track appointment in the biology department at Indiana University-Bloomington, before moving to an appointment as professor and chair of the biology department at Boston College in 2000. His transition from research in developmental genetics into research on malaria biology during 2007 led to affiliations with the Broad Institute as an associate researcher and as an adjunct faculty member at the Harvard School of Public Health.

He joined Biogen in 2013 to build up epigenetics research. The primary goals of the Epigenetics Group are to identify, prioritize and prosecute epigenetic targets for disease indications, with an emphasis on Neurology, and to provide domain expertise in epigenetic biology for all groups within the company. The group's research focuses on epigenetic regulatory mechanisms underlying synaptogenesis and synaptic resilience, on the epigenomics of AD and PD for the discovery of disease mechanism, therapeutic targets and biomarkers of disease onset and progression, and on epigenomic engineering. The Epigenetics Group is utilizing these approaches to garner new insights into epigenetic biology in healthy and diseased cell states, and to leverage those insights for the development of new therapies that will ameliorate or prevent degenerative disease.

Dr Karin Oien

Clinical Senior Lecturer and Honorary Consultant in Pathology,
Institute of Cancer Sciences, University of Glasgow

Karin Oien is Clinical Senior Lecturer in Pathology in the Institute of Cancer Sciences in the University of Glasgow. Dr Oien is also Honorary Consultant in Liver and Gastro-intestinal Pathology, based in the new Queen Elizabeth University Hospital, Glasgow.

Dr Oien's research focusses on molecular pathology and stratified medicine for cancer. She has a long-standing interest in cancer of unknown primary (CUP) and has contributed to national and international CUP guidelines and the UK national CUP-ONE clinical trial, funded by Cancer Research UK (CR-UK), for which Dr Harpreet Wasan is chief investigator. She is also part of the Glasgow team working on pancreatico-biliary cancer, with Professors Andrew Biankin, Jeff Evans, Owen Sansom and colleagues leading in the Wolfson Wohl Cancer Research Centre and Beatson Institute.

Dr Oien has led the Glasgow clinical hub contribution to the CR-UK Stratified Medicine Programme (SMP). She currently leads the Glasgow Molecular Pathology (GMP) Node, within the new Molecular Pathology network funded by the Medical Research Council (MRC) and Engineering & Physical Sciences Research Council (EPSRC). The GMP Node aims to create a dynamic platform that integrates laboratory medicine, including pathology, and informatics disciplines, which handle and analyse the large datasets which emerge from molecular research. Scientists, pathologists and clinicians will work together to develop and perform new tests, and interpret, deliver and act on results for patient benefit. The vision is to develop new tests to better diagnose patients and guide their best treatment in cancer, inflammatory diseases and in cardiovascular and metabolic diseases.

Most recently, Dr Oien has been appointed Chair of the new Cellular and Molecular Pathology (CM-Path) initiative funded by the National Cancer Research Institute (NCRI). Working together with partner organisations, CM-Path aims to re-invigorate UK academic pathology.

Professor Mary Porteous

Mary has been a Consultant Clinical Geneticist in Edinburgh since 1992 and is currently Service lead for the SE Scotland Genetic Service. She holds honorary Professorships at Edinburgh and Edinburgh Napier Universities. She has a longstanding interest in genetic service development, in particular translating research findings into clinical practice. She was the first chair of the Scottish Clin-ical Genetic Forum and chaired the Scottish Molecular Pathology Review Group co-authoring the report which lead to the establishment of a Nationally funded Molecular Pathology Consortium. Mary was a co-founder of the Scottish Genetic Education Network (ScotGEN) and is currently lead examiner for the Certificate of Medical Genetics run through the Royal College of Pathologists.

John Savill

Professor Sir John Savill, BA, MBChB, PhD, FRCP, FRCPE, FRCSEd(Hon), FASN, F.MedSci, FRSE, FRS
Vice Principal & Head of College of Medicine and Veterinary Medicine
Professor of Experimental Medicine

John Savill graduated in Physiological Sciences from Oxford in 1978 and in Medicine from Sheffield in 1981. He received a PhD (London) in 1989. After junior hospital appointments in Sheffield, Nottingham and London, he spent seven years in the Department of Medicine at the Royal Postgraduate Medical School, Hammersmith Hospital, with spells as an MRC Clinical Training Fellow and Wellcome Trust Senior Clinical Research Fellow.

In 1993, he moved to the Chair of Medicine at Nottingham; subsequently moving in 1998 to Edinburgh as Professor of Medicine where he set up and became the first Director of the University of Edinburgh/Medical Research Council Centre for Inflammation Research.

In 2002, he became the first Vice-Principal and Head of the College of Medicine and Veterinary Medicine; he moved to the Chair of Experimental Medicine in 2006.

From 1st June 2008 to 30th September 2010, he was Chief Scientist for the Scottish Government Health Directorates (part-time).

On 1st October 2010 he was appointed as Chief Executive and Deputy Chair of the Medical Research Council, combining this with Head of College duties in Edinburgh.

His work has been recognised by fellowships of the Royal Colleges of Physicians of London and Edinburgh, the Academy of Medical Sciences, the American Society of Nephrology, The Royal Society of Edinburgh, The Royal Society and an honorary fellowship of the Royal College of Surgeons of Edinburgh. He was a member of the Medical Research Council from 2002 to 2008 and chaired two Research Boards during this period. He was knighted in the 2008 New Year's Honours List for services to clinical science.

Professor Colin Semple

A critical component of normal biological function is the elegantly coordinated expression of our genes, reflected in the shifting constellations of thousands of active genes within millions of cells across time and space. The genomes of complex organisms have evolved a huge variety of strategies and systems to reliably achieve this fine scale regulation of gene expression. My group uses computational approaches to understand these regulatory systems in the human genome, how they are disrupted in diseases such as cancers, and how they have evolved.

My PhD was in population genetics at the University of Edinburgh (1994), followed by postdoctoral stints at the University of Michigan and Trinity College Dublin exploring the first genome sequences derived from yeast and worms. In 1998 I joined the MRC Human Genetics Unit, studying the initial human genome sequence to understand human disease predisposition. Since 2001 I have led the Bioinformatics Analysis Core at the MRC Institute of Genetics and Molecular Medicine (IGMM), one of the largest UK MRC research establishments supporting approximately 500 scientists. We provide computational collaborative expertise to IGMM experimental research groups, and also large research consortia such as the Scottish Genomes Partnership. I am also a member of the Edinburgh-St Andrews MRC Molecular Pathology Hub, the EpiGeneSys EU-wide network of excellence in epigenetics and systems biology, various MRC review panels and journal editorial boards.

Dr Clare Turnbull

Dr Clare Turnbull is the Clinical Lead for the Genomics England 100,000 Genomes Cancer Program. Particular focus within this role includes (i) optimising NHS pathology pipelines for cancer sample collection and preparation (ii) interpretation of germline and tumour genomic data for clinical reporting and (iii) engagement of the UK clinical academic community into the program to optimise patient recruitment and research outputs.

She also leads a research team at the Institute of Cancer Research, London, focused on translational studies in cancer genomics and genetic predisposition. Her main research focus has been on the genetic basis of testicular cancer and she has also published extensively on genetic susceptibility to breast, ovarian and childhood cancers. Having trained as a Clinical Geneticist, her clinical work at Guys Hospital NHS Trust focuses on management of patients and families with genetic susceptibility to cancer.

Clare undertook her preclinical training in Cambridge and qualified in medicine from Oxford University. She undertook general medical training across hospitals in Oxford and London and specialist training in Clinical Genetics in London. She completed a PhD in Genetic Epidemiology and Molecular Genetics at the Institute of Cancer Research, London and a Masters degree in Epidemiology and Statistics at the London School of Hygiene.

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