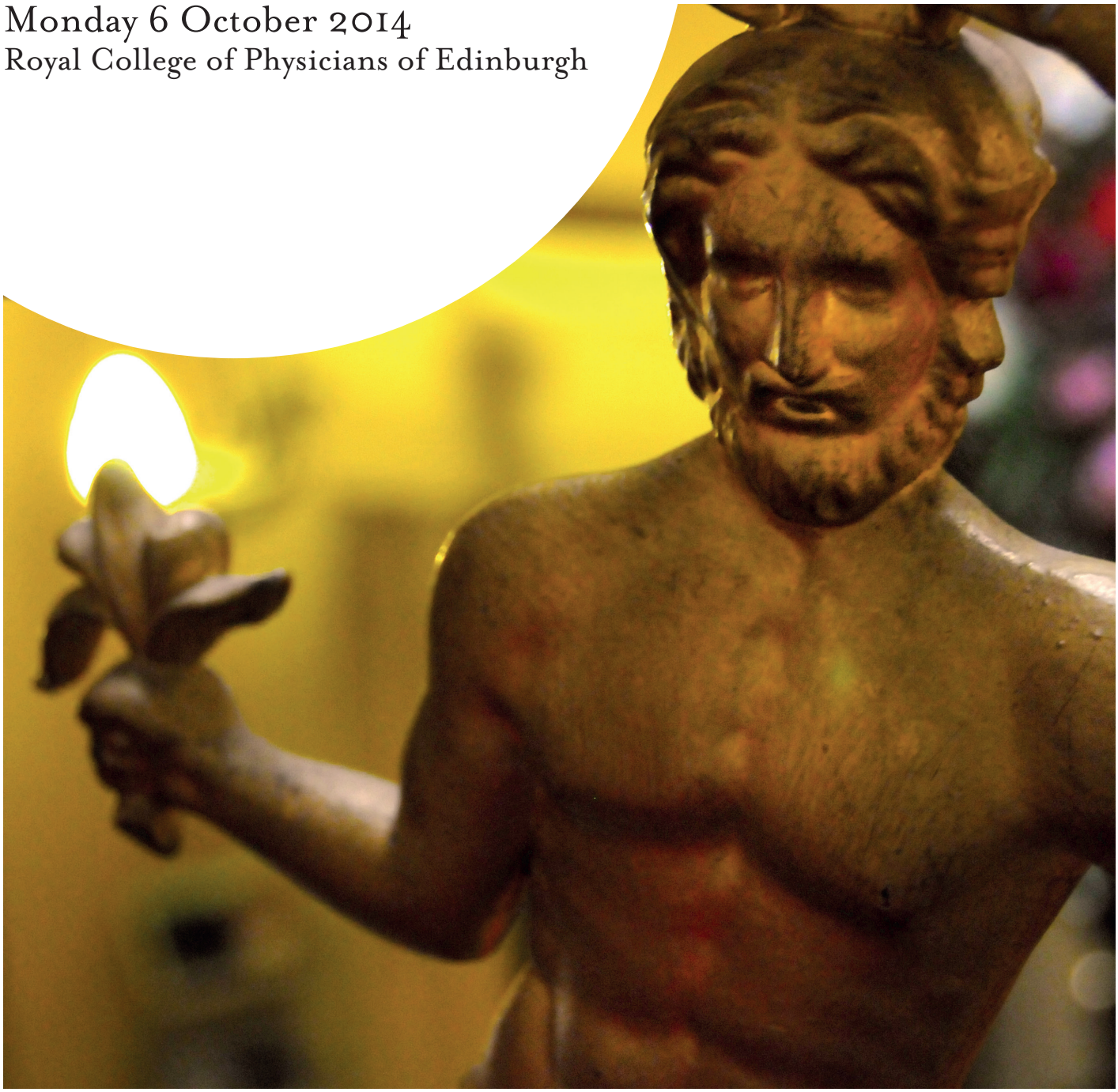
CLINICAL GENOMES SCOTLAND

Monday 6 October 2014
Royal College of Physicians of Edinburgh



THE UNIVERSITY OF EDINBURGH

igmm
INSTITUTE OF GENETICS & MOLECULAR MEDICINE

NHS Lothian

CLINICAL GENOMES SCOTLAND

This meeting will address the question of how the clinical community should deal with the increasing availability of genome sequencing, how Scotland will rise to the challenge raised by the UK 100,000 genomes project, and how genome technology and information can best be applied to healthcare.

Organised by:
Tim Aitman, Nick Hastie, Mary Porteous
The University of Edinburgh and NHS Lothian.

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PROGRAMME

6 October 2014
Royal College of Physicians of Edinburgh

9.00am COFFEE AND REGISTRATION

9.30am **SESSION 1 - Rare Diseases**
Chair - Mary Porteous (Edinburgh)

Han Brunner (Nijmegen). **Genome or Exome Sequencing for Intellectual Disability. What is the Difference?**

Caroline Wright (Sanger Institute) and David FitzPatrick (Edinburgh). **Diagnostic Analysis of Genome-Wide Data in DDD**

Mark Caulfield (Genomics England). **Genomic Medicine - Time for Translation**

10.45am COFFEE

11.15am **SESSION 2 - Common Diseases**
Chair - Anna Dominiczak (Glasgow)

Sean Grimmond (Glasgow). **Mining Oncogenic Drivers and Clinical Utility from Cancer Genomes**

John Todd (Cambridge). **Translating Genetics via Experimental Medicine to Therapy**

Sharon Peacock (Cambridge). **Advances in Translational Microbiology**

12.30pm **KEYNOTE LECTURE 1**
Howard Jacob (Medical College of Wisconsin). **Genomic Sequencing in the Clinic: where is the Utility?**
Chair - Tim Aitman (Edinburgh and Imperial College).

1.00pm LUNCH

1.45pm **KEYNOTE LECTURE 2**
Sir John Bell (Oxford). **Stratified Medicine: Bringing Genomics to the Clinic**
Chair - Nick Hastie (Edinburgh)

2.15pm **SESSION 3 - Clinical Genomes and Commercial Platforms**
Chair - Mark Blaxter (AFFILIATION HERE)

Robert C. Green (Harvard). **Empirical Studies on the Path to Genomic Medicine**

Tim Harris (Biogen). **Exome Sequencing of ALS Patient Genomes and other Stories**

David Bentley (Illumina). **Genomes for Medicine**

Ron A. Andrews (President, Genetic Sciences, Thermo Fisher). **Using NextGen Sequencing to Change the Course of Cancer Management**

3.55pm TEA

4.25pm **SESSION 4 - Bioinformatics**
Chair - David Porteous (Edinburgh)

Janet Thornton (EBI). **The challenges of interpreting genome data**

Tim Hubbard (Kings College). **Data, Annotation and Clinical Feedback for the 100,000 Genomes Project**

5.15 - 5.45pm **KEYNOTE LECTURE 3**
David Goldstein (Duke University School of Medicine). **Toward Precision Medicine in Neurodevelopmental Disease**
Chair - Andrew Morris (Edinburgh)

5.45pm Concluding Comments: **Tim Aitman**

5.50 - 6.30pm Drinks Reception, RCPE

7.00pm CONFERENCE DINNER
(by advance registration only)

Tim Aitman

Professor Tim Aitman is Chair of Molecular Pathology and Genetics at the University of Edinburgh and Consultant Physician in NHS Lothian. He plays a key role in strategic development of the Centre for Genomic and Experimental Medicine and the MRC Institute of Genetics and Molecular Medicine within the University's School of Molecular, Genetic and Population Health Sciences. He is Director Elect of the Centre for Genomic and Experimental Medicine.

A graduate of the Birmingham Medical School and Kings College London he obtained his DPhil at Wolfson College in Oxford. Before joining the University of Edinburgh in April 2014, he was Group Head and Section Chair at the MRC Clinical Sciences Centre, Hammersmith Hospital in London, Honorary Consultant Physician at Hammersmith Hospitals NHS Trust and Professor of Clinical & Molecular Genetics at the Faculty of Medicine of the Imperial College London (where he is still Visiting Professor).

Professor Aitman is a Fellow of the Royal College of Physicians, the Academy of Medical Sciences and the Society of Biology. He is also a Trustee of the Public Health Genomics (PHG) Foundation, a member of several external advisory boards (including the Sir Jules Thorn Medical Advisory Committee and scientific advisory board of the Faculty 1000 Biology), and editorial board member of Mammalian Genome, Physiological Genomics, BMC Bioinformatics and BMC Medical Genomics. He was the Specialist Adviser for the House of Lords Science and Technology Committee's Inquiry into Genomic Medicine, and is currently a member of the Genomics Advisory Board of Health Education England.

Professor Aitman has authored more than 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. His research uses genome technology and information to gain insights into the pathogenesis of both rare and common diseases, and more recently to advance methods for disease diagnosis and stratification.

He has co-ordinated multiple scientific projects and research consortia with career grant support of over £30 million. In 2007, with the support of Nature Genetics and the Wellcome Trust, he co-founded the "Genomics of Common Diseases" meetings, now a prominent international meeting series. Professor Aitman has also been a mentor of many successful graduate students and postdoctoral scientists.

Ron A. Andrews

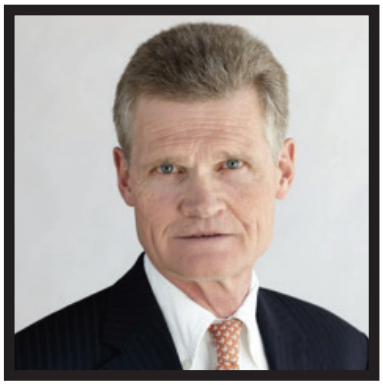
Ron A. Andrews has more than 25 years of experience in the diagnostics industry. He served as the President of Medical Sciences at Life Technologies Corporation from February 2, 2012 to June 26, 2013. In June 2013, Mr. Andrews assumed the role of President of Genetic Sciences, Life Solutions Group, Thermo Fisher Scientific. In his current role he is steward of their $2M Genetic Technology Division and oversees the execution of Thermo Fisher's deployment of their world leading genetic platforms into various applied markets including oncology, inherited diseases, animal health and human identification.

Mr. Andrews served as the Chief Executive Officer of Clarient Inc. (formerly Chromavision Medical Systems Inc.) since July 22, 2004. He served as Chief Executive Officer of GE Molecular Diagnostics. Mr. Andrews served as the President of Clarient Inc. from July 22, 2004 to April 2008. From August 2002, he served as Senior Vice President Global Marketing and Commercial Business Development at Pleasanton, California-based Roche Molecular Diagnostics, where he developed and led the strategic execution for all diagnostic commercial operations. Mr. Andrews was also responsible for executive direction of all marketing functions, directed the development of the 10-year Strategic Plan for the organization, and completed the reorganization of commercial operations during that period.

From 2000 to 2002, Mr. Andrews held two senior executive positions with Indianapolis-based Roche Diagnostics Corporation. He served as Vice President of U.S. Commercial Operations at Molecular Diagnostics and directed sales, marketing, technical field support, and product development activities and was responsible for U.S. commercial strategy development for the clinical laboratory market. He served as Vice President of Marketing at U.S. Commercial Operations and was responsible for planning and directing all aspects of the Roche U.S. Laboratory Systems Commercial Operations Marketing which included the clinical chemistry, immunochemistry, hematology, near patient testing and molecular markets.

From 1995 to 2000 Mr. Andrews served as Vice President of marketing of Atlanta-based Immucor Inc. where he helped lead the transition of that company from a reagent manufacturer to an instrument systems company. Prior to Immucor, he spent almost 10 years in management positions of increasing responsibility at Chicago-based Abbott Diagnostics, culminating in the position of Senior Marketing Manager, Business Unit Operations. He has been Vice Chairman of Clarient Inc. since April 28, 2008 and has been its Director since July 22, 2004. He serves as a Member of Advisory Board at Definiens AG. He serves as Member of Scientific Advisory Board at BioTrove, Inc. He served as Director of Cardiac Science Corporation from November 5, 2009 to December 1, 2010.

Mr. Andrews holds a BS degree in Biology and Chemistry from Spartanburg, SC-based Wofford College in 1981 and has participated extensively in the executive development programs at both Roche and Abbott Labs.



Sir John Bell

Professor Sir John Bell FRS is Regius Professor of Medicine at Oxford University, and Chairman of the Office for the Strategic Coordination of Health Research. He served as President of the Academy of Medical Sciences from 2006 to 2011. As a Rhodes Scholar (1975-78), Sir John undertook his medical training in the UK and then went on to Stanford University, returning to the UK in 1987.

His research interests are in the area of autoimmune disease and immunology where he has contributed to the understanding of immune activation in a range of autoimmune diseases. In 1993, he founded the Wellcome Trust Centre for Human Genetics, one of the world's leading centres for complex trait common disease genetics. In 2001, he was appointed non-executive director of Roche Holding AG and in 2008 he joined the Gates Foundation Global Health Advisory Board which he has chaired since 2012. Sir John was responsible for the working party that produced the highly influential Academy of Medical Sciences "Strengthening Clinical Research" report that highlighted the need for the UK to focus some of its attention on developing expertise in translational research. In December 2011, Sir John was appointed one of two UK Life Sciences Champions by the Prime Minister. He sits on the board of Genome England Limited and chairs its Science Advisory Committee.

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 DPhil 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.

Han Brunner

Han Brunner is currently full Professor and head of the department of Human Genetics at Nijmegen University Hospital, and at Maastricht University Medical Center. He has initiated and conducted several research projects that use clinical genetic observations as the starting point for human molecular genetic investigations into such topics as intellectual disability, human behaviour, skeletal development, brain development, neuromuscular disease, congenital malformations, and gonadal development and function.

By pioneering the innovative use of novel genome analysis tools, and their successful application to intellectual disability research, Han Brunner has shown that small submicroscopic chromosome and de novo gene mutations may well be the most important causes of ID. This de novo paradigm now allows the development of accurate diagnostic strategies for this group of patients. He has published more than 300 peer-reviewed manuscripts, which have collectively attracted > 15,000 citations. His H-index is currently 80.

HONOURS

2013: Elected member of the Royal Netherlands Academy of Sciences

2013: Knight in the order of the Dutch Lion

2012: Elected member of Academia Europea.

2012: Ernst Klenk lecture of the Center for Molecular Medicine Cologne, Germany.

2012: Stanley Davidson Lecture, Royal College of Physicians, Edinburgh, Edinburgh UK.

2011: Radboud Science Award (with Dr Joris Veltman), Radboud University Nijmegen.

2009: Frank Greenberg memorial lectureship, Baylor College of Medicine, Houston USA.

1995: Ben ter Haar prize of the Clinical Genetics Society of the Netherlands.

1994: Prize of the the Dutch Organisation for Research of Neuromuscular diseases, for research by young investigator.

- President European Society of Human Genetics, 2013-2014
- Member of the Board of Directors American Society Human Genetics, 2013-2016
- Member Executive Board European Society of Human Genetics, 2012-2015
- Chairman Scientific Program Committee European Society of Human Genetics 2003-2010
- Co-chairman Diagnostics, International Rare Diseases Research Consortium (IRDiRC), 2012-present
- Member of the Scientific Advisory Board of the Sydney Brenner Institute of Molecular Biology, Johannesburg, South Africa, 2011-present
- Organizer of the European School of Genetic Medicine with Professors G. Romeo (Bologna), B. Wirth (Cologne), 2007-present

KEY PUBLICATIONS

1. Vissers et al, 2010. A de novo paradigm for mental retardation. Nat Genet. 42:1109-1112.
2. de Ligt et al, 2012. Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. N Engl J Med. 367:1921-1929.
3. Gilissen et al, 2014. Genome sequencing identifies major causes of severe intellectual disability. Nature 511:344-347.

Mark Caulfield

Mark graduated in Medicine in 1984 from the London Hospital Medical College and trained in Clinical Pharmacology at St Bartholomew's Hospital (Barts) where he developed a research programme in molecular genetics of hypertension and clinical research. In 2009 he won the Lily Prize of the British Pharmacology Society. He is a Fellow of The Royal College of Physicians.

In 2000 Mark successfully bid for £3.1m to create the Barts and The London Genome Centre at the Queen Mary University of London which now underpins over 40 programmes of research. Since 2008 he has directed the Barts National Institute of Health Research (NIHR) Cardiovascular Biomedical Research Unit. In 2012 he become Co-Chair of the NIHR Comprehensive Research Network Cardiovascular Sub-Speciality Group.

Mark was appointed Director of the William Harvey Research Institute in 2002 and was elected to the Academy of Medical Sciences in 2008. His particular areas of research are Cardiovascular Genomics and Translational Cardiovascular Research and Pharmacology.

From 2009 to 2011 Mark was President of the British Hypertension Society. He has also served on the NICE Guideline Group for hypertension and leads the Joint UK Societies' Working Group and Consensus on Renal Denervation which he has driven from research into NHS care.

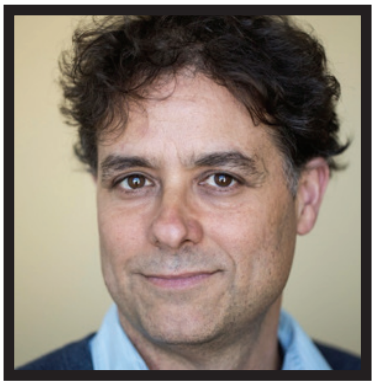
In 2013 he became an NIHR Senior Investigator and was also appointed as the Chief Scientist for Genomics England (NHS 100K Sequencing Project) 2013-2015.

David R FitzPatrick MD, FRCP, FRCPath

David FitzPatrick is an academic paediatric geneticist based in at the MRC Human Genetics Unit where he leads a lab-based research group focused on understanding the genetic basis of developmental disorder and is joint head of the Medical and Developmental Genetics Section. He is a graduate of the University of Edinburgh Medical School. He trained in paediatrics and clinical genetics the Royal Hospitals for Sick Children in Edinburgh, Bristol and Glasgow. He then became a Wellcome Trust Clinical Training Fellow at the University of Glasgow and subsequently a Howard Hughes Clinical Research Training Fellow with Professor David Valle at Johns Hopkins Hospital in Baltimore. He returned to Edinburgh as a consultant in Clinical Genetics in 1994 and moved to the MRC HGU in March 2000.

He has a long standing research interest in the genetic causes of major eye malformations, and in close collaboration with Professor Veronica van Heyningen the Edinburgh teams have identified the two major genetics causes of anophthalmia (heterozygous, de novo, loss-of-function mutations in either SOX2 and OTX2). This work has been rapidly translated into clinical diagnostic practice throughout the world. The current focus is on identifying the causes of severe bilateral coloboma and the use of whole exome sequencing has enabled the identification of several novel loci such as YAP1 and MAB21L2. A common theme of this work is the requirement for innovative uses of genetic technologies to identify and validate causative mutations affecting the structure or regulation of individual genes.

David FitzPatrick was one of six applicants on the large-scale, UK-wide, trio-based exome sequencing project Deciphering Developmental Disorders (DDD) that was funded by the Health Innovation Challenge Fund in 2010. He is a member of the management committee of DDD and leads both on the development of the DDG2P database which is used as an important component of the reporting pipeline and the analysis to identify discriminative phenotypic features that predict particular disease genes.



David Goldstein

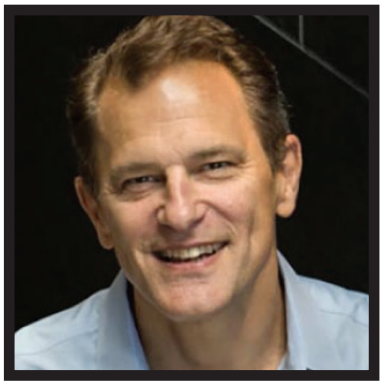
Dr. Goldstein was trained in theoretical population genetics and has studied many aspects of human genetic variation over the last 20 years with a particular focus on the genetics of disease and treatment response. His early work focused primarily on genetic anthropology, while his more recent work has focused primarily on medical genetics and pharmacogenetics. Dr. Goldstein was Professor of Genetics at University College London from 1999 - 2005. In 2005 he moved to Duke University as a Professor in the Departments of Molecular Genetics and Microbiology and Biology, and the Director of the Center for Human Genome Variation (CHGV.) In 2010 he was appointed the Richard and Pat Johnson Distinguished University Professor.

Under Dr. Goldstein's leadership, the CHGV has emerged as a leading human genetics research center with a number of seminal discoveries, including de novo mutations in ATP1A3, the gene responsible for Alternating Hemiplegia of Childhood, the role of IL28B in treatment response to Hepatitis C infection, and was a leader in the field of demonstrating the potential of next generation sequencing in diagnosing rare genetic and neurological conditions. Dr. Goldstein is also a principal investigator of Epi4K, the NINDS Epilepsy genetics Center without Walls and he directs its genome sequencing and bioinformatic core. Epi4K is currently the largest epilepsy genetics project in the world and is in the process of generating whole exome and whole genome sequence data on no less than 4,000 patients with epilepsy.

Dr. Goldstein has authored over 200 scholarly publications in the areas of population and medical genetics. He received the Royal Society/Wolfson research merit awards in the UK for work in human population genetics, the Triangle Business Journal Health Care Heroes Award: Innovator/Researcher Award (2008), the University of North Carolina Clinical Services Award (2012). He chaired the Gordon Research Conference in Human Genetics & Genomics (2013) and currently serves on the NIH-NINDS Advisory Council, and was elected a fellow of AAAS in 2013.

KEY PUBLICATIONS

1. Enns GM, Shashi V, Bainbridge M, , ….. Goldstein DB. Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway. Genet Med. 2014 doi: 10.1038/gim.2014.22. PMID: 24651605
2. Epi4K & EPGP Investigators. De novo mutation in the classic epileptic encephalopathies. Nature 2013, Sep 12;501(7466):217-21 doi: 10.1038/nature12439. Epub 2013 PMID:23934111, PMCID: PMC3773011.
3. Heinzen EL, Swoboda KJ, Hitomi Y, , ……., Goldstein DB. De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. Nat Genet 2012, 44(9):1030-4. PMID: 22842232, PMCID: PMC3442240.



Robert C. Green

Robert C. Green, MD, MPH is a medical geneticist and physician-scientist who directs the G2P Research Program (genomes2people.org) in translational genomics and health outcomes in the Division of Genetics at Brigham and Women's Hospital and Harvard Medical School.

Dr Green is principal investigator of the NIH-funded REVEAL Study, in which a cross-disciplinary team conducted 4 separate multi-center randomized clinical trials since 2000, collectively enrolling 1100 individuals in order to explore emerging themes in translational genomics. Dr Green also co-directs the NIH-funded PGen Study, one of the first prospective studies of direct-to-consumer genetic testing services. He is principal investigator of the MedSeq Project, the first NIH-funded randomized trial to explore the use of whole genome sequencing in the clinical practice of medicine and co-directs the BabySeq Project, the first NIH-funded trial of sequencing in newborns. The MedSeq and BabySeq Projects utilize genome sequencing both in patients who are affected with hereditary disease and in those who are healthy, in order to study downstream impact on health, behavior and health care costs.

Dr. Green is currently Associate Director for Research of the Partners Personalized Medicine, a Board Member of the Council for Responsible Genetics and a member of the Informed Cohort Oversight Boards for both the Children's Hospital Boston Gene Partnership Program and the Coriell Personalized Medicine Collaborative. He was lead author of the recently published recommendations from the American College of Medical Genetics and Genomics for management of incidental findings in clinical sequencing.

Sean Grimmond

Professor Grimmond originally trained in Biochemistry and obtained a Ph.D. in Human Genetics from the University of Queensland's Department of Pathology (1991-1994). He completed postdoctoral studies at the Queensland Institute for Medical Research (1994-1996) and the MRC Mammalian Genetics Unit & Mouse Genome Centre in Harwell (1997-1999). He returned to Australia in 2000 and established the Institute for Molecular Bioscience's Expression genomics laboratory. In 2009 he was promoted to Professor, became the founding Director of the Queensland Centre for Medical Genomics and was awarded Australia's ICGC Cancer Genome Program (2009-2014). In 2011 he became a Founding Fellow of the Royal College of Pathologists of Australasia's Faculty of Science. In September of 2013 he moved to the University of Glasgow and took up the new chair in Medical Genomics.

Over the last 20 years, his research has focused on surveying genomes & transcriptomes and discerning the underlying genetics controlling biological processes and pathological states. His scientific achievements are broad and include: i) Cloning and commercialization of the Vaso-endothelial Growth Factor VEGFB (1995-97), ii) Contributing to the positional cloning of the MEN1 tumour suppressor gene (1991-97), iii) Defining the functional diversity of mammalian transcription in the FANTOM consortia (2002-07), iv) Pioneering transciptome and genome technologies (microarrays, massive scale in-situ hybridization, RNAseq, WES, WGS & WTS), v) Diagnosing novel neurodevelopmental disorders via NGS (2011-13), vii) Discerning the basis and evolution of antibiotic resistance (2012-13). He is best known for sequencing the first Australian Cancer patients in 2010 and resolving the molecular basis of pancreatic, ovarian and oesophageal cancer through large scale cohort WGS sequencing (2009-), Professor Grimmond has been awarded the Australian Eppendorf Medal for Genomics (2003), Julian-Wells medal for Transcriptomics (2010), NH&MRC Research Fellowship (2007-2011), NH&MRC Principal Research Fellowship (2012-2014) and Royal Society Wolfson Merit award (2013-2017). He has published more than 180 articles, >11,000 citations and holds 5 patents.

Tim Harris

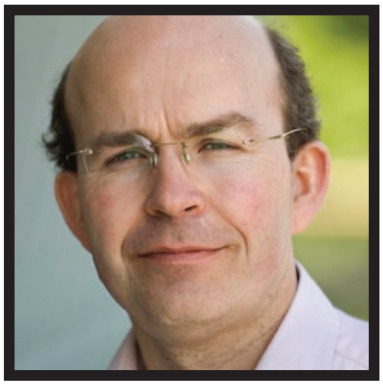
Dr Tim Harris is a science and business leader with over 35 years of experience guiding and leading laboratory work and scientists in a range of research technologies. He is a molecular biologist and biochemist, and currently serves as the SVP for Translational Sciences and Technology at Biogen Idec. He was the Chief Technology Officer (CTO) and Director of the Advanced Technology Program (ATP) at SAIC-Frederick, Inc. in Maryland which operates the National Cancer Institute's leading center for cancer and AIDS research (now Frederick National Laboratory operated by Leidos Inc.,). He has served as President and Chief Executive Officer (CEO) of Novasite Pharmaceuticals. He founded SGX Pharmaceuticals in 1999 (formerly Structural Genomix), where he built the company to more than 130 employees, raised $85M in capital, and generated more than $20M in revenue during six years as CEO before it was sold to Eli Lilly. Before founding SGX, Dr. Harris was Senior Vice President, Research and Development at Sequana/Axys. He began his career working on animal viruses (at the Institute of Animal Health in Surrey) such as that causing Foot & Mouth Disease and was one of the first molecular biologists at Celltech (now UCB Pharma) in the UK. He subsequently spent five years at Glaxo Group Research as Director of Biotechnology from 1989 to 1993.

Nick Hastie

Professor Nick Hastie is Director of the MRC Human Genetics Unit and the Institute of Genetics and Molecular Medicine in Edinburgh. Nick Hastie was born in North Wales, where he attended Colwyn Bay Grammar School. He went on to receive an honours degree in Medical Microbiology at Liverpool University. Following that, he carried out his PhD work on Influenza Replication at Cambridge University. Since then, he has worked in many areas including gene expression, genome organisation (including telomeres) and protein evolution. He has had a long-term interest in human developmental genetics, concentrating on the childhood kidney cancer, Wilms' tumour. Over the past few years Professor Hastie has also become heavily engaged in a major population genetics project to identify genetic risk factors for common disease.

Nick Hastie was an International Scholar of the Howard Hughes Medical Institute; he chaired the General Motors Cancer MOT Awards Committee and sat on the General Motors Assembly. He is Chairperson of a number of Scientific Advisory Boards, including (until 2010) that for the Sanger Institute and the Wellcome Trust Centre for Human Genetics. He was European Editor of Genes & Development for a decade and currently sits on the Advisory Boards of Genes & Development and PLoS Biology. Professor Hastie is a Fellow of the Royal Society, a Fellow of the Royal Society of Edinburgh, a Member of EMBO and was awarded a CBE for Services to Science in 2006. He was awarded the Medal of the Genetics Society of the UK in 2008.

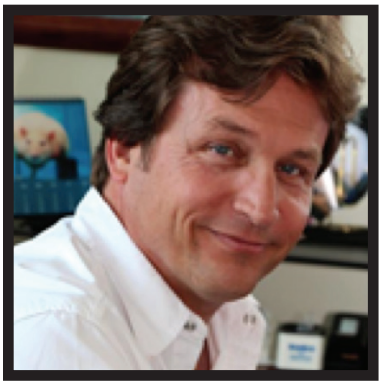
In his spare time Nick likes to do body combat, sing, listen to music, read, garden and watch movies and rubbish on television.



Tim Hubbard

Professor Tim Hubbard is Head of the Department of Medical & Molecular Genetics at King's College London and Director of Bioinformatics for King's Health Partners. He is also Head of Bioinformatics at Genomics England, the company set up by the Department of Health to execute the 100,000 genomes project. Until 2013 he was Head of Informatics at the Wellcome Trust Sanger Institute where he remains Honorary Faculty. While at Sanger he was one of the organisers of the sequencing of the human genome. In 1999 he co-founded the Ensembl project to analyse, organise and provide access to the human genome and from 2007 led the GENCODE project to annotate the structure of all human genes.

He is actively involved in efforts to improve data sharing in science, develop open access publishing resources and plan for the adoption of genomic medicine. He is a member of the cross funding agency Expert Advisory Board on Data Access (EAGDA) and is chair of the advisory board of Europe PubMedCentral. He was a member of the OSCHR e-health board, which advised on the use of electronic patient record data for research, supporting the work to create the Clinical Practice Research Datalink (CPRD) and the Farr Institute. He was a member of the Bioinformatics and Education, Engagement and Training working groups of the Human Genomic Strategy Group (HGSG) and a member of the Scottish Health Informatics Research Advisory Group (HIRAG). He was seconded part time to NHS England as specialist advisor before joining Genomics England.



Howard J. Jacob, PhD

Warren P. Knowles Chair of Genetics
Director, Human and Molecular Genetics Center
Professor, Departments of Physiology and Pediatrics
Medical College of Wisconsin

Professor Jacob received his Ph.D. in Pharmacology from the University of Iowa in 1989. He completed two parallel post-doctoral fellowships in functional genomics and molecular genetics/genomics at Harvard, Stanford and MIT with Victor J. Dzau, M.D. and Eric S. Lander, Ph.D. He was on the faculty at Massachusetts General Hospital and Harvard Medical School for nearly 4 years before moving to Milwaukee. In 1996, he joined the Medical College of Wisconsin as an Associate Professor, Department of Physiology with full Professorship and Tenure in 2001. He was appointed the Founding Director of the Human and Molecular Genetics Center (HMGC) and was awarded the Warren P. Knowles Chair of Genetics in 1999.

Under his leadership the Human and Molecular Genetics Center (HMGC) has grown from two faculty members to 30 and is one of the top funded genetic programs based on NIH funding. Dr Jacob has over 240 peer reviewed papers. His work over the last 25 years has focused on finding genes responsible for common complex diseases.

In 2010 Dr. Jacob led a team of researchers at the Medical College who used an innovative DNA sequencing technique to unravel the medical mystery of Nicholas Volker, a young boy whose life-threatening disease had baffled his doctors and tested his family's faith. Working with Medical College scientists and physicians at the Children's Hospital of Wisconsin, Dr. Jacob's team used Nicholas' DNA to diagnose his disease and recommend a course of treatment. This treatment has so far been successful.

Dr. Jacob and his team have developed a full end-to-end Genomics Medicine Clinic for Children and Adults. They conduct gene tests, gene panel tests, whole exome sequencing and whole genome sequencing in a CAP/CLIA laboratory.

Sharon Peacock

Sharon Peacock is Professor of Clinical Microbiology and an Honorary Faculty Member at the Wellcome Trust Sanger Institute. Prior to returning to the UK in 2009, Professor Peacock spent 7 years in Thailand where she was head of bacterial diseases research at the Wellcome Trust Major Overseas Programme in Bangkok, and directed a programme of clinical and basic research into melioidosis and Burkholderia pseudomallei. Her UK research focus is the translation of rapid, high-throughput microbial sequencing into diagnostic and public health microbiology. She receives funding from the Medical Research Council through a UKCRC (UK Clinical Research Collaboration) Translational Infection Research Initiative Consortium Grant, and from the Department of Health and Wellcome Trust through a Health Innovation Challenge Fund Award. Professor Peacock is deputy chair of the Medical Research Council Infection and Immunity Board and undertakes a range of other peer review activities for the MRC. She contributed to the UK Chief Medical Officer's Annual Report (published 2013) on antimicrobial resistance, led the working group on scientific priorities for the infectious diseases component of the Department of Health 100,000 genome project, and is a member of the Public Health England implementation committee for infectious diseases sequencing. Professor Peacock is a Fellow of the Academy of Medical Sciences.

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 Clinical 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 has participated in a wide range of collaborative research projects including the cloning of the 2 major genes involved in Hereditary Haemorrhagic Telangiectasia with Doug Marchuk and 2 CRUK Programme grants held with Malcolm Dunlop and Harry Campbell on the genetic contribution to Colorectal Cancer. She participates in a range of multicentre clinical studies including the International Huntington's Disease registry study ENROLL HD.

As chair of the Royal College of Physicians SAC training committee for Medical Genetic Trainees, Mary was responsible for the delivery of the 2010 curriculum. She is currently lead examiner for the Certificate of Medical Genetics run through the Royal College of Pathologists.

Janet Thornton

Since 2001 Janet has been Director of the EMBL-European Bioinformatics Institute on the Wellcome Trust Genome Campus at Hinxton, near Cambridge, UK. EMBL-EBI provides core bioinformatics services, especially public biological data for genome and protein sequences and structures, small molecule data and integrated proteomic, metabolomics and transcriptome information. The institute also performs investigator led bioinformatics research.

In addition Janet is an EMBL research group leader and the goal of her research is to understand biological processes at the molecular level, by studying protein structures and sequences using computational approaches. Her work involves the classification of protein families and structures to elucidate the principles governing their folding and evolution with a recent focus on protein variants and their effects on function. Her group is also involved in studies to understand the molecular basis of ageing.

Janet coordinated the ELIXIR preparatory phase (2006 – 2013) and leads the commission-funded BioMedBridges project, which clusters the biomolecular sciences research infrastructures to provide data bridges and ensure data interoperability between them.

Janet has worked on various government committees to address the bioinformatics and training challenges of genomic medicine.

John Todd

John Todd FRS, FMedSci, PhD is Professor of Medical Genetics at Cambridge University and Director of the JDRF/Wellcome Trust Diabetes and Inflammation Laboratory (DIL) in the University's Cambridge Institute for Medical Research. Todd researches type 1 diabetes (T1D) genetics and disease mechanisms with an aim of clinical intervention. Previously, Todd was Professor of Human Genetics at Oxford University and a Wellcome Trust Principal Research Fellow.

Todd helped pioneer genome-wide genetic studies, first in mice and then in humans. He then went on to study the associations between mapped genomic disease-associated regions and phenotypes by founding and deploying the Cambridge BioResource. In the latest phase of his research, to translate basic genetic and immunological knowledge to treatment and prevention, the DIL has now completed its first mechanistic, statistically adaptive, drug dose-finding trial in T1D patients, establishing new and effective methods of trial design, governance, conduct and patient recruitment. The DIL has a major role in training and mentoring others in patient/people-based research, promoting data/sample access and sharing, and can advise other laboratories and industrial partners in immunotherapeutics and experimental medicine.

Caroline Wright

Dr Caroline Wright works at the Wellcome Trust Sanger Institute in Cambridge, where she manages the Deciphering Developmental Disorders project, a UK-wide translational research study that uses genome sequencing to investigate the genetic causes of severe undiagnosed developmental disorders. She was formerly Head of Science at the Foundation for Genomics and Population Health, where she remains a Fellow, and is also a member of the personal genomics blog GenomesUnzipped. She trained at the University of Cambridge, where she studied natural sciences followed by a PhD in biophysics.

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