Faculty profiles

1. Professor Dhavendra Kumar, Medical Director/ CEO, Genomic Medicine Foundation (d.kumar@qmul.ac.uk; genomicmedicine@yahoo.co.uk)



Professor Dhavendra Kumar, MBBS, MD, DCH (RCPSI), MMedSci, PGCertMedEd, FRCPI, FRCP, FRCPCH, FACMG, DSc.(Hon)
The William Harvey Research Institute, Bart's and The London School of Medicine & Dentistry, Queen Mary University of London, UK

Academic Affiliations:

- *Hon. Consultant in Clinical Genetic/ Cardiovascular Genetics, Inherited Cardiac Unit, Bart's Heart Centre, St. Bart's Hospital, Bart's NHS Foundation Trust, London, UK*
- *Consultant in Clinical Genetics & Genomic Medicine, Cardiff Spire Hospital, Cardiff, Wales, UK*
- *Senior Consultant Adviser, 'Apollo Genomics Institutes', Apollo Group of Hospitals, India.*
- *Hon. Professor of Medicine, Swansea University, Wales, UK*
- *Visiting Professor, Centre for Health Genomics, University of South Wales, UK*
- *Visiting Professor, Centre for Genomic Healthcare, University of South Wales, Pontypridd, Wales, UK*
- *Senior Faculty Adviser to the 'Centre of Precision Medicine & Health', King George Medical University, Lucknow, India*
- *Hon. Professor, Department of Medical Genetics and Genetic Counselling, JSS Academy of Higher Education (Deemed University), Mysore, India.*
- *Hon. Professor, Department of Biotechnology, Shri Mata Vashno Devi University, Katra, Jammu, India*
- *Hon. Professor, Department of Medical Genetics, Kasturba Medical College, Manipal, Karnataka, India.*
- *Visiting Professor, Centre for Advanced Research & Education, Chettinad Medical College & Health University, Chennai, Tamil Nadu, India.*
- *Honorary Visiting Professor, Faculty of Medicine, University of Colombo, Sri Lanka*
- *Visiting Professor, School of Medicine, Texas Tech University, Texas, USA*
- *External Adviser for Genomic Medicine, South Africa Medical Research Council (SAMRC), South Africa*
- *Honorary Adjunct Professor, Mahatma Gandhi University Medical Sciences Technology, Jaipur, India*

Citation: A highly acclaimed, globally acknowledged genetic and genomic clinician with special interests in clinical genetics, cardiovascular genomics and genomic/ OMIC medicine. He is credited with landmark contributions and achievements in genetic-inherited diseases of children, hereditary familial conditions of heart and blood vessels, applications of novel genomic principles and technology in genomic-precision medicine and public and population health genomics. He is honoured with ***Doctor of Science, Honoris Causa*** by the alma mater, KGMU Lucknow, ***Hind Rattan*** International NRI Award, the ***GAPIO-Siemens Medical Innovation Award***, the ***Glory of Georgians & Life Time Achievement Award*** of the King George's Medical University Alumni UK, and the **Life Time Achievement Award conferred by GAPIO (2023).** He is widely applauded for his sincere and persistent efforts for medical genetics as the integral part of medical teaching and practice across India and in low and middle income countries (LMICs).

Notable services, achievements and contributions:

- **Author/Editor** of several peer reviewed research papers, articles and books. Main book titles include- ***Genetic Disorders of the Indian Subcontinent (Kluwer-Springer); Genomics and Clinical Medicine (Oxford); Principles and Practice of Genomic Medicine (Oxford); Principles and Practice of Clinical Cardiovascular Genetics (Oxford); Cardiovascular Genetics & Genomics- Principles and Clinical Practice (Springer-Nature); Oxford Specialist Handbook Inherited Cardiac Disease (Oxford); Medical & Health Genomics (Elsevier); Genomics and Society (Elsevier); Clinical Molecular Medicine-Principles & Practice (Elsevier); Genomic and Molecular Medicine e-book series (9 volumes), Morgan& Claypool Publishers. Advances in Genetics series (9 volumes), Academic Press by Elsevier.***

2. Professor Michael Hubank



Dr Mike Hubank

Scientific Director, Clinical Genomics, Royal Marsden Hospital, London
Scientific Director, NHS England North Thames Genomic Laboratory Hub
Professor of Translational Genomics, Institute for Cancer Research, London

Biography

Dr Hubank has worked in Genomics for over 20 years. Following a PhD at UCL, he worked at the University of Sussex and Yale University before returning to the UCL Institute of Child Health in 2000 to found and run *UCL Genomics*. In 2016 he moved into clinical diagnostics at the Royal Marsden Hospital, London, to lead a translational laboratory focused on the development and clinical application of genomic assays for cancer diagnostics. Since 2018 he has been Scientific Director at the NHS England North Thames Genomic Laboratory Hub.

Research Interests

Mike is a joint investigator on several research projects using multiomic profiling for optimising personalised diagnostics and treatments for childhood cancer patients. He has a strong focus on developing and applying genomic assays, and a particular interest in moving circulating biomarkers into standard of care testing.

@generoom

Contact: Michael.hubank@icr.ac.uk

3. Professor Thomas Connor, School of Biosciences, Cardiff University



The research that is undertaken in Professor Connor lab seeks to understand pathogen variation in order to answer a range of questions from how pathogens evolve, to how and why they spread in local and global outbreaks. This work has a number of labels, but broadly speaking we use genomic epidemiology, phylogenomics and population genetics approaches to answer our research questions.

Prof. Connor's research is underpinned by whole genome sequencing, using the data from organisms genomes to work out how they are related to, and different from, other organisms of interest. This work is heavily computational with mathematical and computational approaches to analyse and interpret the "Big Biological Data". In a practical sense this work has potential for clinical applications in diagnostic and surveillance services in the NHS in Wales for HIV, TB and other Mycobacteria, C. difficile and Influenza.

The Connor group is part of the recently formed Microbiomes, Microbes and Informatics (MMI) group (webpage underdevelopment). The MMI group currently comprises the research groups of Thomas Connor, Esh Mahenthiralingam, Julian Marchesi and Andrew Weightman, and has over 25 active research staff and postgraduate students.The MMI group are highly research active generating over £3.5 million in grant income between 2010 and 2017, and publishing extensively in top journals (cumulative h index > 150, > 400 publications, and > 25,000 citations; source Scopus.com).

Roles

- Module Leader: BI3252 The 'omics revolution (Bioinformatics & Functional Genomics)
- Biocomputing Research Hub lead
- Member of College of Biomedical and Life Sciences Data Strategy Group
- Member of Supercomputing Wales Infrastructure Committee
- Member of Cardiff Supercomputing Facility Oversight Group
- Wales regional lead and technical lead for the Cloud Infrastructure for Microbial Bioinformatics
- Bioinformatics Lead for the Public Health Wales Pathogen Genomics Unit.

4. **Professor Michael Barnes**



Professor Michael Barnes, Professor of Bioinformatics and Director of the Centre for Translational Bioinformatics, William Harvey Research Institute, Queen Mary University of London.
m.r.barnes@qmul.ac.uk

Michael co-leads the Centre for Translational Bioinformatics (C4TB), at Queen Mary University of London and is a Fellow of the Digitial Environment Research Institute, QMUL. His team work across diverse research areas, including genomics, drug discovery, stratified medicine, machine learning and health informatics with a unified objective to drive forward translation into the clinic. He brings an industrial perspective to the C4TB, drawn from 16 years of leadership of bioinformatics teams in the pharmaceutical industry.

Michael is a Fellow of the Alan Turing Institute and an HDR-UK Investigator, and co-leads the Genomics England Stratified medicine genomic interpretation clinical partnership. He has an active portfolio of stratified medicine projects as a co- investigator and data integration lead on several MRC projects, including MRC PSORT (Psoriasis), MRC RA-Map (RA), MRC MATURA (RA), and CLUSTER (Juvenile Arthritis). He also co-leads the NIHR AI-Multiply Consortium which is using AI to investigate the relationship between multiple long-term conditions and polypharmacy. He has served on the MRC Methodology Research Panel and Stratified Medicine panels and also advises on a number of project boards, including the UnitedHealth Group Pharmacogenetics advisory group, the Dutch Heart Foundation, the MRC-eMedLab HPC Cloud facility, the IMI etriks project, and the F1000 faculty.

5. Prof. Matt Brown, Chief Scientist, Genomics England & King's College, London



Chief Scientific Officer, Genomics England; Professor of Medicine, King's College, London

Professor Matthew Brown, an internationally renowned clinician-scientist, joined Genomics England in 2021.

Prior to joining Genomics England he was Director of the National Institute for Health Research (NIHR) Guy's and St Thomas' Biomedical Research Centre and Professor of Medicine within the Faculty of Life Sciences and Medicine, King's College London.

Professor Brown trained as a clinician-scientist and a rheumatologist in Australia and the UK. Previous positions include Professor of Musculoskeletal Sciences at the University of Oxford; Director of the Australian Translational Genomics Centre, Distinguished Professor, and Director of Genomics at the Queensland University of Technology; and Director of The University of Queensland Diamantina Institute and Professor of Immunogenetics at The University of Queensland.

He has made contributions to the development of gene-mapping approaches in human diseases and genome-wide association study methodology, leading to the discovery of thousands of genetic variants, with a particular interest in ankylosing spondylitis, rheumatoid arthritis, and osteoporosis.

In the genetics of rare human diseases, he has identified genes responsible for monogenic forms of arthritis, ectopic bone development, and skeletal dysplasias. He has also led efforts in Australia to translate research sequencing capability into precision medicine programs for cancer patients.

Professor Brown was elected a Fellow of the Australian Academy of Sciences in recognition for his achievements in genetics research. He still practises medicine, in the specialty of rheumatology, with a particular focus on axial spondyloarthropathies.

6. Professor Claudia Langenberg, William Harvey Research Institute, Queen Mary University of London (claudia.langenbrg@qmul.ac.uk



Claudia is the Director of PHURI and the former programme Leader at the MRC Epidemiology Unit at the University of Cambridge.

Claudia is a public health specialist combining her expertise with research focused on molecular epidemiology. Her work integrates large-scale genomic and metabolomic data to discover, prioritise and characterize metabolic pathways and test their causal relevance and specificity across a range of diseases, uncovering genetic influences on thousands of molecules in the blood, using this knowledge to better understand human disease such as type 2 diabetes.

In 2018 Claudia received the Helmholtz International Fellow Award and was named in the top 20 female scientists in the UK in 2022.

7. Professor Sir Munir Pirmohamed, University of Liverpool, UK

Professor- Pharmacology and Therapeutics & Sid David Weatherall Professor of Medicine (munirp@liverpool.ac.uk)



Professor Sir Munir Pirmohamed (MB ChB, PhD, FRCPE, FRCP, FFPM. FRSB, FBPhS, FMedSci) is David Weatherall Chair in Medicine at the University of Liverpool, NHS Chair of Pharmacogenetics, and a Consultant Physician at the Royal Liverpool University Hospital. He is Director of the Centre for Drug Safety Sciences, and Director of the Wolfson Centre for Personalised Medicine. He is also Director of HDR North. He is an inaugural NIHR Senior Investigator, Fellow of the Academy of Medical Sciences in the UK, Commissioner on Human Medicines. He was President of British Pharmacological Society from January 2020-December 2021, and is currently President of the Association of Physicians. He was awarded a Knights Bachelor in the Queen's Birthday Honours in 2015.

His research focuses on personalised medicine, clinical pharmacology and drug safety in a variety of disease areas, including cardiovascular medicine.

8. Professor Sir Mark Caulfield



Professor Sir Mark Caulfield is Professor of Clinical Pharmacology at Queen Mary University of London and the Vice Principal for Health for Queen Mary's Faculty of Medicine and Dentistry.

Professor Caulfield graduated in Medicine in 1984 from the London Hospital Medical College and trained in Clinical Pharmacology at St Bartholomew's Hospital, he developed a research programme in molecular genetics of hypertension and translational clinical research.

At Queen Mary University of London Professor Caulfield has made contributions to the discovery of genes related to blood pressure, cardiovascular health, cancer and rare diseases. His research has changed national and international guidance for high blood pressure.

He has won the Lily Prize of the British Pharmacology Society, the Bjorn Folkow Award of the European Society of Hypertension 2016 and the Franz Volhard Award of the International Society of Hypertension in 2018.

Professor Caulfield was appointed Chief Scientist for Genomics England in 2013, charged with delivery of the 100,000 Genomes Project on whole genome sequencing in rare disease, cancer and infection. He was instrumental in delivering the 100,000 Genomes Project which has delivered life-changing results for many patients. He worked with NHS England to co-create the National Genomic Test Directory, which offers equitable access for 56 million people to appropriate genomic tests. Professor Caulfield was awarded a knighthood in 2019 for his leadership of the 100,000 Genomes Project.

He is a member of the Barts Health NHS Trust Board, the Barking, Havering and Redbridge University Hospitals NHS Trust, the MedCity Board and is the President Elect of the British Pharmacological Society

9. Dr Hywell Willaims, Senior Lecturer Bioinformatics, Division of Cancer & Genetics, Cardiff University (WilliamsHJ1@cardiff.ac.uk)



Dr. Williams's research is focused on improving our understanding of the biological systems that when dysregulated lead to disease phenotypes in humans. Primarily he is working on rare diseases with an emphasis on neuro-related phenotypes.

One of his main interests is how gene expression is regulated both spatially and temporally during development and throughout life and how DNA variation can influence this. To understand this will require knowledge of how the genome is organised in 3D space, which regions of the genome are in contact to regulate gene expression and the full repertoire of variation each individual possesses within both the coding and non-coding genome.

His aim is to improve our ability to make a diagnosis in the 50-60 % patients with rare disease who currently remain undiagnosed and armed with this knowledge build a better understanding of the functional biology that leads to disease. In turn this will improve our ability to develop new therapeutics to treat patients and in the future may even allow us to prevent these diseases form occurring.

To do this will require the use of techniques such as spatial transcriptomics, Hi-C and whole genome sequencing. The analysis of the Big Data generated will require smart algorithms and informatics to extract the maximum amount of information in a meaningful way. He believes that network analysis techniques will be hugely important in helping us combine this information so we understand the composition of biological systems in healthy and disease states.

To achieve our goals will require working together as a community to share our data in an open and transparent way.

10. Dr. Tootie Bueser, Chief Nurse, North Thames Genomic Medicine Service; Director of Nursing and Midwifery Southeast GMSA. (tootie.beuser@nhs.net)



Dr. Tootie Bueser is a Health Education England/National Institute for Health Research Clinical Doctoral Research Fellow at the Florence Nightingale Faculty of Nursing, Midwifery and Palliative Care. Her work is focused on developing a new intervention to improve psychosocial and educational support for patients who have a new diagnosis and/or carrier status for an inherited cardiac condition.

Tootie Bueser is an experienced cardiac nurse and is the lead nurse for the Inherited Cardiac Conditions services at King's College Hospital and Guy's and St Thomas' Hospital.

She is the President of the British Association for Nursing in Cardiovascular Care and is thereby a Council member of the British Cardiovascular Society and the UK representative for the European Society for Cardiology Association of Cardiovascular Nurses and Allied Professionals. She is an active member of the Association for Inherited Cardiac Conditions; and the Cardiovascular and the Ethics & Social Sciences Genomics England Clinical Interpretation Partnership for the 100,000 Genomes Project. Tootie Bueser is on the editorial board of the British Journal of Cardiac Nursing.

Tootie Bueser is a member of the Clinical Expert Advisory Group for Cardiomyopathy UK and volunteers for their patient support line.

11. Prof. Helen Firth, Director, DECIPHER, Sanger Genome Centre, Hinxton, Cambridge, England; Newham College, University of Cambrdige (hvf21@cam.ac.uk)



Professor Helen Firth, DM, FRCP,FMedSci is a Consultant Clinical Geneticist at Cambridge University Hospitals. Her main research interests are in mapping of the clinical genome and the matching of rare genomic variants to empower discovery and diagnosis in rare disease.

She is an Honorary Faculty Member of the Wellcome Trust Sanger Institute, an Honorary Professor of Clinical Genomics: at Addenbrooke's Hospital in the Department of Medical Genetics and a Bye-Fellow of Newnham College. Helen chairs the Joint Committee on Genomic Medicine of the Royal College of Physicians, Royal College of Pathologists and British Society of Genetic Medicine.
After the human genome was sequenced in 2004, she initiated the DECIPHER project (http://decipher.sanger.ac.uk) to put that data to use. DECIPHER enables clinicians and scientists around the world to share information about rare genomic variants to facilitate diagnosis and help to elucidate the role of genes whose function is not yet known. More than 280 centres of clinical genetics around the world now contribute data to DECIPHER and the project has been cited by >2,200 publications in the peer-reviewed scientific literature. In June 2020, a major update to DECIPHER (v.10) now allows researchers and clinicians to input, interpret and share all types of genomic variants in all regions of the genome. DECIPHER today continues to bring emerging knowledge of human genetics and genomics to the forefront of clinical practice.
https://www.sanger.ac.uk/news_item/update-to-decipher-brings-power-to-clinical-genomics/
In 2020, Helen was elected as a Fellow of the Academy of Medical Sciences.

Research Interests

Since 2010 Professor Firth has been Clinical Lead for the Deciphering Developmental Disorders study (DDD study) (http://www.ddduk.org) one of the world's largest nationwide, genome-wide sequencing projects in rare disease. The DDD study is a partnership project between the UK NHS and Wellcome Trust Sanger Institute that is using detailed genomic analysis of ~13,500 children with severe developmental disorders to improve the diagnosis of these conditions and to further the understanding of their genomic architecture and biology. The DDD study has now enabled diagnoses for thousands of children across the UK and has resulted in the discovery of >50 new genes and in >200 publications in the scientific literature.

13. Professor Patricia Munroe, Queen Mary University of London, UK
(p.b.munroe@qmul.ac.uk)



Professor Patricia Munroe (Patsy) graduated with a B.Sc. in Biochemistry, and M.Sc. in Biotechnology from the University of Galway, Ireland. She then worked at the Wellcome Trust Research Laboratories for six months before commencing a PhD in cardiovascular genetics at St Bartholomew's Hospital. Patricia was awarded a PhD in Medicine in 1995 and following successful post-doctoral fellowships at University College London (NIH funded), she joined the William Harvey Research Institute as a Lecturer in 1998. In 2007 she was appointed Professor of Molecular Medicine.

Prof Munroe's lab investigates the molecular basis of cardiac arrythmia's, hypertension and heart failure. Our research includes genomic studies of cardiovascular risk factors as a route for elucidating disease mechanisms, the development of 'omic biomarkers and clinical models for improved risk prediction, pharmacogenetics and personalised medicine.

She co-leads several international complex genetic disease consortia. Using meta-analysis of genome-wide association studies (GWAS) and large-scale candidate gene studies she has discovered over 1000 genetic loci associated with hypertension. She has also identified hundreds of genetic loci for the electrocardiogram and cardiac magnetic resonance measures of heart structure and function. . As a member of the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium she co-leads projects leading to the discovery of loci for electrocardiogram markers and gene x environment interactions for cardiovascular risk factors. She is the Director of the Genome Centre at FMD and an Adjunct Professor at the Department of Physiology and Pharmacology, The University of Toledo, USA.

She was listed as a Highly Cited Researcher by Thomson Reuters in 2015, 2016, 2017 and 2018 (Top 1% in Molecular Biology & Genetics). She was elected as a Fellow of Academy of Medical Sciences in 2021.

14. Professor Panos Deloukas, Director, WHRI, Queen Mary University of London, UK (p.deloukas@qmul.ac.uk)



Panos Deloukas obtained his BSc in Chemistry from the Aristotelian University of Thessaloniki, Greece and MSc in Microbiology from University Paris 7, France. He received his PhD from the Biozentrum University of Basel, Switzerland in 1991. He joined the Sanger Centre in 1994 where he set up a high-throughput pipeline for radiation hybrid mapping, leading an effort to map 30,000 gene markers, GeneMap98. Panos was an active member of the Human Genome Project coordinating the sequencing and analysis of chromosomes 10 and 20. After the completion of the HGP he joined the International HapMap project constructing SNP maps of the human genome. Since 2005 he is studying the molecular basis of common disease and variable response to drugs in humans through large-scale genetic studies. He joined the William Harvey Research Institute at Queen Mary University London in September 2013 working on the genomics of coronary artery disease and lipid levels. Panos is a member of many consortia including CARDIoGRAMplusC4D, Global Lipids Genetics Consortium, GIANT, the UK Biobank Cardiometabolic Consortium, and the Cardiovascular Genomics England Clinical Interpretation Partnership. He has authored over 400 publications (H-index 121) and is listed by Thomson Reuters among the 1% highly cited researchers in Molecular Biology & Genetics since 2012**.**

15. Professor Dame Sue Hill, Chief Scientific Officer, NHS Genomic Medicine, NHS England.



Professor Dame Sue Hill DBE FMedSci FRSB FRCP(Hon) FRCPath (Hon) FHCS (Hon) is the Chief Scientific Officer (CSO) for England and a respiratory scientist by background. Throughout her career she has led on large-scale priority programmes across government and in NHS England including as the senior responsible officer for Genomics in the NHS, introducing a world-leading and nationwide Genomic Medicine Service, building on her work in heading up the NHS contribution to the 100,000 Genomes Project.

She has also played a pivotal role in the national COVID-19 programme leading the development and deployment of testing technologies into use for the UK population and co-directing the whole-genome sequencing of SARS-CoV-2 programme.