Faculty profiles

1. Professor Dhavendra Kumar



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 Thomas Connor, Cardiff University



Professor Thomas Connor , School of Biosciences

The research that is undertaken in my 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.

Historically we have examined questions on a range of pathogens, including gastrointestinal pathogens (E. coli, Salmonella, Shigella, C. difficile) which collectively account for over a billion cases of disease around the world every year, pathogens which are particularly associated with antimicrobial resistance and viruses including HIV and Influenza.

My 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, and we use and develop mathematical and computational approaches to analyse and interpret the "Big Biological Data" that we and our collaborators generate.

This work is also readily translatable, and I have spent the past few years working closely with colleagues in the NHS to translate the approaches we develop into diagnostic and surveillance tools that can be used at a local and national level. In a practical sense this has seen us take approaches we use in our resarch, and build clinical diagnostic and surveillance services in the NHS in Wales for HIV, TB and other Mycobacteria, C. difficile and Influenza. Our Pathogen Genomics work is undertaken with the Public Health Wales Pathogen Genomics Unit and Genomics Partnership Wales. In addition to my research team, I also lead the Bioinformatics team within the Pathogen Genomics Unit.

Our HIV work was highlighted in the 2020 Cardiff University Innovation Awards: https://www.cardiff.ac.uk/business/why-work-with-us/who-we-work-with/healthcare-innovation

As part of the COVID-19 pandemic, almost all of my time has been spent supporting the pandemic response in Wales. We have taken our expertise in other pathogens to develop the capacity to sequence and analyse COVID-19 from Welsh patients, sequencing more than 7,500 SARS-CoV-2 genomes since the start of the pandemic. We use this data to understand the spread of the pandemic over time, and to support outbreak response and national surveillance efforts. This work is undertaken as part of the COVID-19 Genomics UK Consortium.

In addition to my pathogen research activities, I have an additional focus on the development and design of research infrastructures to support the analysis of the genomic datasets that we produce. This work has included the design and development of national computational resources in the UK, as well as the design of computational resources used in translational settings in the NHS and Industry. The largest of these infrastructures, MRC CLIMB, which I helped design and play a role in leading supports over 1,000 microbiology researchers from across the UK, and has been the key peice of UK infrastructure that has underpinned the COG-UK sequencing effort to date. The CLIMB-COVID system has provided a core infrastructure for the collation and analysis of over 70,000 SARS-CoV-2 genomes sequenced across the UK.

Microbiomes, Microbes and Informatics

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).

The four current MMI staff recently moved (June 2017) to a single shared location within a new £1.6 million refurbished area of the Sir Martin Evans Building. This comprises a large class II certified research laboratory, equipment and tissue culture rooms, a group office area and academic offices. The MMI group welcomes approaches by potential fellowship applicants and funded PhD students to host their research and expand our strategic research on Microbiomes, Microbes and Informatics.

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.

3. Dr. James Timmons, Queen Mary University London



Reader- Translational Bioinformatics, Queen Mary University London, UK

James graduated from the Universities of Glasgow and Nottingham (Physiology and Pharmacology) and then spent 7yrs as a team-leader in the pharmaceutical industry (responsible for *in vivo* and *in vitro* models). He ran a team working on lead identification and leading optimisation for metabolic disease, thrombosis and atherosclerosis. He set up one of the first human clinical studies (1998) to use global transcriptomics to stratify human responses to exercise therapy and led the pharmacology for an anti-thrombotic drug nomination (2002).

He moved to the Karolinska Institute in 2003 to re-train in RNA biology, particularly noncoding RNA. During this time his group discovered, with the Cannon lab, the developmental link between 'brown' adipocytes and muscle. He was appointed to Chair in Exercise Physiology (2006), first at Heriot-Watt and later at Loughborough University (2013) and a 5yr visiting Professorship at University of Stockholm (2007). In Edinburgh, his group discovered that very brief (<5min) intermittent high intensity exercise ('HIT') was sufficient to improve insulin action, challenging 50 years of exercise advice (work that featured in a BBC Horizon documentary). In 2011, he led a FP7 consortium that validated the principal of HIT in randomised clinical trials, and in 2019 WHO exercise guidelines were altered to reflect the work from several independent groups. As Director of Research, he led the School of Biological Sciences RAE2008 return.

James has also held Professor posts (Research) at the Royal Veterinary College and King's College London – running molecular physiology studies - funded by the MRC, BBSRC, NIH and Industry – while managing his EU FP7 multi-centred HIT trial. His group have used machine learning to build the first molecular classifier for cardiorespiratory adaptability in humans (2010), and the first transcriptomic multi-tissue classifier of human age (2015). In 2017, after two decades of 'wet-lab' activities, he decided to focus full time on bioinformatics - joining WHRI in 2021, as a Senior Fellow (Reader in Data Science). From 1998 to 2022 James has trained >20 graduate and post-doctoral scientists in industry and academia. He has an H-index of 49 (from 90, mostly first or senior authored, articles). He is a visiting Professor at the University of Miami, and a member of the Royal Society of Medicine and the Biochemical Society.

4**. Prof. Matt Brown, Chief Scientist, Genomics England & 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.

5. Prof. Maik Pietzner, Precision Healthcare Unit, QMUL, London, UK



Chair in Health Data Modelling

As the newly appointed chair for Health Data Modelling, Maik has a keen interest in the computational integration of different health data modalities to translate (big) data into better health for patients, with a particular focus on underrepresented diseases and patient groups. He co-leads the group for Computational Medicine at the Berlin Institute of Health, Germany, focusing on the discovery of new genetic variants that predispose to so far neglected diseases that he and his team integrate with large-scale molecular data sets to identify novel drug targets or opportunities to repurpose already existing drugs. Maik has an outstanding track record with publications in Science and the Nature family.

6. Professor Sir Munir Pirmohamed, University of Liverpool, 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.

7. 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.

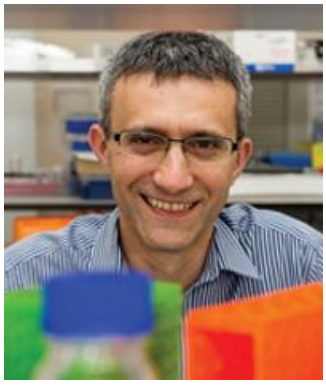
At Queen Mary University of London Professor Caulfield has made contributions to the discovery of genes related to blood pressure, cardiovascular health, cancer and rare diseases. His research has changed national and international guidance for high blood pressure.

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

8. Professor Ed Tobias, University of Glasgow, Scotland



Prof Edward Tobias is an educator and researcher in medical genetics and genomics. He is also an (honorary) consultant clinical geneticist in the NHS.

Personal qualifications: BSc (1st class Hons), MBChB, MRCP, PhD (Molecular Biology), FRCP, FHEA, FAoP (Assocn. of Physicians).

He is the recipient of the Education Award 2021 of the European Society of Human Genetics and an invited education sub-committee lead of the global Human Genome Organisation (HUGO International).

Research

He currently supervises post-graduate research (into developmental genes) involving whole-genome DNA sequencing or RNA analysis. He is Chief Investigator of a whole-exome and whole-genome sequencing study, a co-investigator on a £6m CSO/MRC-funded WGS Scottish Genomes Partnership and a collaborator with investigators at the Wellcome Trust Sanger Institute, Cambridge and at the Francis Crick Insitute, London.

Previously, with a personal MRC Fellowship award, he undertook a PhD in the molecular biology of receptor signalling, followed by post-doctoral research in molecular cancer genetics & bioinformatics. After subsequent training in NHS Clinical Genetics, he won a GSK Senior Clinical Research Fellowship, funding his tumour suppressor gene research, publishing results in Nature Genetics.

Education

Also passionate about teaching & training, & having previously gained Fellowship of the UK Higher Education Academy, he is lead author of international textbooks, chapters, educational websites and smartphone apps, used worldwide. He is Clinical Director of the MSc in Medical Genetics & Genomics (which has now won UK, Scottish & university awards) as well as the MSc in Genetic and Genomic Counselling. He is also lead for a masters-level cutting-edge practical Clinical Genomics course (with >60 Masters students in 2021) and for multi-award-winning undergraduate MBChB year 3 Genetics teaching. He was joint lead of the training workstrand of the £3.4m MRC/EPSRC Glasgow Molecular Pathology Node & joint Clinical Director of the MSc in Molecular Pathology.

He co-created a 3D molecule viewer virtual reality app for the education of bio-medical students, which was a finalist for the UK Technology & Innovation T.H.E. Award 2020. He is also a member of a (award-winning) STEM public engagement team and won a teaching award following voting by a class of 300 final-year medical students (May 2018).

He is an invited sub-committee lead of the Human Genome Organisation International Education Committee and a member of the European Society of Human Genetics (ESHG) Education Committee and Scottish Genetics Education Network (ScotGEN) Steering Committee. He is also an invited active participant in the Clinical Genetics and Genomics examination steering committee of the European Union of Medical Specialists. His educational genetics and genomics website (EuroGEMS.org) has been adopted by the ESHG and by HUGO-International as their principal online education source and has already been used in over 130 countries.

He is also a lead educator for two University of Glasgow Massive Open Online Courses (MOOCs) and an Expert Track MOOC on the FutureLearn online platform.

He felt greatly honoured to be awarded the prestigious ESHG Education Award at their 2021 conference. The award was last made in 2017.

He enjoys receiving genetics and genomics questions from students, on any course.

He is enormously grateful to all of his colleagues in the university (including, in particular, the Medical Genetics & Genomics team) and in the NHS, past and present, for all of their invaluable assistance and support.

9. Dr. Tootie Bueser, Guy's and St Thomas' NHS Foundation Trust, London, UK



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.

10. Prof. Helen Firth, Director, DECIPHER, Sanger Genome Centre, HInxton, Cambridge, England



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.

11. Professor Patricia Munroe, Queen Mary University of London, 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.

12. Professor Panos Deloukas, Director, WHRI, Queen Mary University of London, 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**.**

13. 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.