9th Annual Open Meeting

Working together to deliver personalised/precision medicine

#UKPGx2022

Attendance 8 CPD Credits

College of Physicians, London

15 June 2022

Welcome to the 9th UK Pharmacogenetics & Stratified Medicine Network Annual Open Meeting

Welcome, we would like to thank all our speakers for kindly taking the time to present their expertise at the meeting. We also thank our sponsors and exhibitors for supporting the event. Finally, we are very pleased to welcome the delegates who are working within academia, industry, healthcare and regulatory sectors to move personalised/precision medicine forward; along with representatives from charities and patient groups supporting research projects. We are privileged to have such a great line up of speakers and hope their presentations will encourage discussion. We are a not-for-profit organisation and all proceeds from the meeting go directly to covering the costs of running our events and maintaining the Network.

The Network is continuing to grow and we have over 1,200 members. You are most welcome to join the Network, membership is free, just contact Christine cjmcn@liverpool.ac.uk. We are at present working on our website and are looking forward to launching a new website shortly. The new website will provide a wealth of information on events, educational / funding opportunities, and organisations delivering personalised medicine into the clinic. Presentations from our events will also be available on the website.

Every year we hold workshops that invite leading experts from all sectors to come together to discuss, and offer solutions to, the challenges of implementing genomic medicine into clinical practice. Future workshops include an Open Targets workshop on 18th October 2022 to discuss the importance of genetics in drug discovery, target prioritisation and therapeutic hypothesis building. A workshop on Decision Support Systems that aid prescribing will be held on 9th November 2022. Please contact Christine cjmcn@liverpool.ac.uk for more details.

Enjoy the day, we hope you are able to network with colleagues from different sectors and develop research collaborations.

Professor Sir Munir Pirmohamed

Programme Overview – Morning

08.00-09.00	Registration and refreshments	
09.00-10.40	SESSION 1: Looking to the Future	Chair: Professor Sir Munir Pirmohamed
09.00-09.10	Welcome	Professor Sir Munir Pirmohamed
09.10-09.40	UK Biobank: a unique combination of scale, depth, duration … and accessibility	Professor Sir Rory Collins UK Biobank
09.40-10.10	Making healthcare personal – the growing role of genomics and microbiome innovation”	Richard Hebdon Innovate UK
10.10-10.40	Can we integrate Pharmacogenetics in Our Future Health?	Professor David Hunter University of Oxford
10.40-11.20	Morning coffee kindly sponsored by MC Diagnostics	
11.20-12.35	SESSION 2: Early career	Chair: Dr Karen Spink
11.20-11.40	Pharmacogenomics in Sub-Saharan Africans	Dr Innocent Asiimwe University of Liverpool
11.40-12.00	Pharmacogenomics implementation – the RCP-BPS report	Dr Emma Magavern Queen Mary University of London
12.00-12.20	Microbiome in drug response: navigating the genetic landscape of the E. coli pangenome	Dr Daniel Martinez MRC London Institute of Medical Sciences
12.20-12.40	Modelling a two-stage screen for autosomal dominant familial hypercholesterolaemia (FH)	Jasmine Gratton University College London
12.40-13.50	Lunch and exhibitor’s stands	

13.15-13.45 During the lunch break there will be a series of informal “discussion tables” to provide cross sector networking opportunities. Please join any table to meet others who share your interests.

Programme Overview – Afternoon

13.50-15.20	**SESSION 3: Genomics** **Chair: Dr Philippa Brice**	
13.50-14.20	**Implementation of pharmacogenomics – results of the UPGx Consortium Pre-emptive Pharmacogenomic Testing for Preventing Adverse Drug Reaction study**	**Professor Henk-Jan Guchelaar Leiden University Medical Centre**
14.20-14.50	**Polygenic risk scores and disease stratification**	**Professor Douglas Easton University of Cambridge.**
14.50-15.20	**Genomics England – looking to the future**	**Professor Matthew Brown Genomics England**
15.20-15.40	**Afternoon tea**	

15.40-17.10	**SESSION 4: Omics Technologies** **Chair: Professor Ann Daly**	
15.40-16.10	**Insights from the metabolome in UK biobank**	**Heli Julkunen Nightingale Health UK Biobank Initiative**
16.10-16.40	**Proteomics in the era of large-scale biobanks**	**Professor Claudia Langenberg Berlin Institute of Health at Charité & University of Cambridge**
16.40-17.10	**Human cell atlas and its relevance to precision medicine**	**Professor Muzlifah Haniffa Newcastle University**
17.10	**Close of conference**	**Professor Sir Munir Pirmohamed**

Speakers are presenting their own research and opinions

PRESENTATION ABSTRACTS

SESSION 1: Looking to the Future

UK Biobank: a unique combination of scale, depth, duration … and accessibility
Professor Sir Rory Collins

UK Biobank is unique because of its combination of very large size (500,000 participants) and very great detail (extensive information about participants' characteristics), complemented by information about their health outcomes from prolonged follow-up through NHS health records … that is, it combines breadth, depth and duration.

The 500,000 participants aged 40-69 years when recruited during 2006-2010 throughout England, Scotland and Wales were identified from National Health Service central registries. After they had given their informed consent (including to access their medical and other health-related records), participants answered detailed questionnaires, had physical measures, and provided blood, urine and saliva samples.

Standard biochemical and haematological assays have been done in all 500,000 participants and all of them have not only been genotyped but also sequenced, with various –omic assays now being started. In addition, 100,000 participants are undergoing detailed imaging studies (including MR of brain, heart and body, and DXA of bones and joints), making it 10 times larger than any previous imaging study conducted anywhere in the world.

UK Biobank is available to both academic or commercial researchers for any health-related research. Since the UK Biobank resource was first made available to researchers in 2012, about 30,000 researchers have registered to use it, with about three-quarters now from outside the UK (In particular, North America and Continental Europe). About 4,500 papers based on UK Biobank had been published by the end of 2021 (about 1700 in 2021 alone), with about 120,000 citations to those papers.

Making healthcare personal :
The growing role of genomics and microbiome innovation"
Richard Hebdon

Innovate UK, is the nation's innovation agency with a mission to help UK businesses to grow through the development and commercialisation of new products, processes and services. Historic investment in the health and life sciences by Innovate UK has been around £2 billion and represents ~25% of our total investment to date. Precision medicine continues to be an important innovation area for Innovate UK support and investment with more than £153.5 million invested in precision medicine and diagnostics innovation between 2007 and 2020. The increasing significance of genomics and microbiome research and innovation and applications in areas such as pharmacogenomics is recognised and will be explored in this presentation.

Can we integrate Pharmacogenetics in Our Future Health?
Professor David Hunter

Pharmacogenetics has conventionally focused on identifying variants in a gene or multiple genes that alter the efficacy or side effect profile of prescription medicines. We were interested in whether polygenic risk scores (PRS) alter the efficacy of commonly used drugs for hypertension and hypercholesterolaemia. We generated PRS for systolic blood pressure and plasma cholesterol in the UK Biobank among participants on hypertensive medications or statins. The PRS for SBP was associated with uncontrolled hypertension among patients on at least one antihypertensive (odds ratio 1.70; 95% confidence interval: 1.60-1.80) top vs. bottom quintile, equivalent to a 5.4 mmHg difference in SBP, and was associated with incident MACE [hazard ratio (HR) 1.13; 1.04-1.23]. The PRS for LDL-C was associated with uncontrolled hypercholesterolaemia (HR 2.78; 2.58-3.00) but was not associated with subsequent MACE. These results suggest that polygenic risk scores can identify patients who may need more frequent monitoring, dose escalation, or combination therapy for these conditions. Polygenic risk scores are a major focus for the Our Future Health project (Ourfuturehealth.org.uk). We plan to enrol up to 5 million adults in the UK over time, and run a genome array with participant consent. We have integrated over 800 known pharmacogenetic variants in the Our Future Health genome array that will be available to approved researchers for research purposes. We are asking participants for consent for re-contact and are exploring ways that PGx information can be made available to participants.

SESSION 2: Early career

Pharmacogenomics in Sub-Saharan Africans
Dr Innocent Asiimwe

Warfarin is the anticoagulant of choice in Sub-Saharan Africa, but as in other populations, dosing is highly variable due to demographic, clinical and genetic factors. Despite increasing acceptance for diversity in pharmacogenomic studies, black African patients remain under-represented in pharmacogenomic research. This is due to reasons that include: lack of a critical mass of African researchers interested in pharmacogenomics, lack of suitable funding and difficulties in recruiting the large sample sizes required for the extremely diverse African populations. This talk describes warfarin anticoagulation in sub-Saharan Africa, highlighting why this remains an area of unmet medical need. Next it explores ethnic diversity and warfarin pharmacogenomics and demonstrates how black African patients remain under-represented in pharmacogenomic studies. The work that we have done in this region, including the development of a clinical warfarin dose-initiation algorithm, the evaluation of machine learning techniques using sub-Saharan African data, and pharmacogenomic studies, is thereafter explored, with the talk concluding with future work/opportunities in the region.

Pharmacogenomics implementation: the RCP-BPS report
Dr Emma Magavern

National Pharmacogenomics implementation poses many exciting challenges. The Royal College of Physicians and British Pharmacological Society jointly led a working group to examine how the NHS could harness the potential of Pharmacogenomics by broad implementation in daily clinical care. Implementation would have to be supported across primary and secondary care settings, funded nationally, and responsive to iterative improvement feedback loops. There would need to be education and decision support tools for clinicians, and meaningful dialogue with the public. Ongoing research in pharmacogenomics must be facilitated.

Microbiome in drug response: navigating the genetic landscape of the E. coli pangenome
Dr Daniel Martinez

Metformin is the first-line therapy for treating type 2 diabetes and a promising anti-aging drug, and it has been demonstrated that the microbiome is intimately linked to drug response in the host. Microbes interact in different ways with xenobiotics by altering their chemical structure and bioactivity or affecting their bioavailability. This variability is generally based on the vast microbial diversity that can be found in the human gut. In fact, the large genetic variability that can be found in a single species, defined as its pangenome, has not been fully explored yet in the context of drug activity. To explore the genetic determinants that drive drug activity, we set up a high-throughput platform where we examined more than 750 different strains of Escherichia coli and their effects in metformin treatment, using the nematode Caenorhabditis elegans as an animal model and biosensor. We observe that host response to metformin is strain dependant, and that it doesn't correlate with microbial growth in the presence of metformin. However, when considering the genetic content within the pangenome, we see that metformin on the host effects can be related to microbial metabolic pathways as ATP synthesis, drug transporters, phosphotransferase system, iron transport or heterologous pathogenic islands. Our high-throughput screening platform paves the way to navigate the complex host-microbe genetic landscape for drug activity, thus improving our ability to predict and act on potential targets in the future.

Modelling a 2-stage screen for monogenic familial hypercholesterolaemia using UK Biobank
Jasmine Gratton

Objective: To evaluate the performance of two-stage adult population screening for autosomal dominant familial hypercholesterolaemia (FH).

Design: We modelled use of different low-density lipoprotein cholesterol (LDL-C) cut-offs (stage 1) to select individuals for DNA sequencing to identify FH-causing variants in LDLR, APOB, APOE and PCSK9 (stage 2), using data from UK Biobank. We also modelled the number of additional FH cases detected by cascade testing of first-degree relatives of index cases and compared this approach with child-parent screening for FH.

Main outcome measures: For different LDL-C cut-offs, we estimated the stage 1 detection and false positive rate, the proportion of individuals requiring sequencing, and the number of FH cases identified by population screening followed by cascade testing.
Results: We identified 488 individuals with an FH-causing variant and 139,951 without (prevalence 1:288). An LDL-C cut-off of >4.8 mmol/L had a stage 1 detection rate (sensitivity) of 40% (95%CI: 36-44%) for a false positive rate of 10% (95%CI: 10-11%). Using this LDL-C cut-off to screen 100,000 individuals (with an estimated 347 FH cases) would generate 10,398 stage 1 screen positives for sequencing, detect 138 FH cases, miss 209, with a further 207 cases being detected through two-generation cascade testing of first-degree relatives of index cases. This is about a third as many FH cases as childhood screening with three generation cascade testing, for twice the sequencing burden.
Conclusions: Two-stage adult population screening for FH could help achieve the FH case detection target in the NHS Long Term Plan, but less efficiently than childhood screening and with a greater sequencing requirement.

SESSION 3: Genomics

Implementation of pharmacogenomics: results of the UPGx Consortium Pre-emptive Pharmacogenomic Testing for Preventing Adverse Drug Reaction (PREPARE) study
Professor Henk-Jan Guchelaar

The benefit of pharmacogenetic testing prior to starting therapy has been well documented for a number of single gene-drug combinations. However, the clinical utility of pre-emptive genotyping of a pharmacogenetic panel has not been assessed.

We conducted an open, multi-center, controlled, cluster-randomized, cross-over implementation study of a pre-emptive 12-gene pharmacogenetic panel in seven EU countries (NCT03093818). Patients (n= 6,944) receiving a first prescription for a drug with a clinical recommendation in the guidelines of the Dutch Pharmacogenetics Working Group (DPWG) were genotyped for 50 germline variants in 12 genes. Patients in the study arm were treated based upon their pharmacogenetic test results according to the DPWG recommendations. Patients in the control arm received standard treatment. The primary outcome was the incidence of causal clinically relevant adverse drug reactions (ADR) within 12 weeks of follow-up. Incidences were compared between the study and control arm. In the presentation, the preliminary results of the PREPARE study will be presented. The U-PGx programme is funded by the European Community's Horizon 2020 Programme under grant agreement no. 668353.

Polygenic risk scores and disease stratification
Professor Douglas Easton FMedSci FRS

Genome-wide association studies have successfully identified thousands of common variants (SNPs) associated with human traits including many cancers and other diseases. Polygenic risk scores (PRS), representing the combined effects of multiple risk SNPs, have been shown be a strong predictor of risk: as a result, there has been considerable interest in utilising PRS to stratify disease risk and improve

the effectiveness of population screening. Theoretical modelling indicates that this should be an effective strategy, but there are many organisational and societal hurdles to overcome before this is practical reality.
Risk scores also show associations with tumour subtypes, but GWAS have so been much less successful in the identification of variants association with tumour behaviour and disease outcome. Risk prediction can be significantly improved by combining PRS with other types of data, including imaging and lifestyle risk factors, and rarer gene variants conferring higher risks of disease, such as those in BRCA1 and BRCA2.
Most GWAS data to date has been generated on individuals of European ancestry, and a major ongoing challenge is to improve risk prediction in individuals of other ancestry and develop risk scores that are generalizable across ancestries. This is critical to ensuring the providing equity of access.

Genomics England – looking to the future
Professor Matthew Brown

Genomics England has commenced a three-year pilot study of whole genome sequencing (WGS) of newborns to improve diagnostic rates of treatable rare diseases, potentially also including pharmacogenomic variants. Should this pilot succeed and WGS be rolled out across the NHS, eventually a point will be reached where all people born in our country will have had their genome sequenced. This could enable amongst other things, pre-emptive pharmacogenomic testing. Analysis of the 100,000 Genomes study has shown that nearly all participants carry at least one CPIC level A variant detectable by WGS, suggesting that use of the WGS data for pre-emptive pharmacogenomic testing is likely to be beneficial. However, not all pharmacogenetic variants are reliably detected by short-read sequencing approaches. Further, different ancestral groups have marked differences in the frequencies of pharmacogenetic variants, and the lower level of genomic study of non-white ancestral groups means that it is likely that they harbour as yet unidentified variants. And of course there are major implementation challenges to overcome to deliver the potential of this approach. Given the cost to our country and high morbidity levels caused by adverse drug reactions though, this would seem like a worthwhile target.

SESSION 4: Omics Technologies

Insights from the metabolome in UK biobank
Heli Julkunen

Blood lipids and metabolites are biomarkers for future disease onset. Many such biomarkers are used in research settings, but only few make it to routine clinical use. This talk will present how detailed metabolic profiling in the UK Biobank provides novel scientific insights and can be translated to consumer applications.

Nightingale Health Plc. has developed a high-throughput metabolomics platform, which quantifies diverse blood biomarkers from multiple metabolic pathways, including lipoprotein measures, fatty acids

and small molecules such as amino acids, ketones, and glycolysis metabolites. This platform has been widely used in cohort studies and trials and has resulted in more than 350 scientific publications. Recently, Nightingale's nuclear magnetic resonance (NMR) platform was used to profile the UK Biobank. The first tranche of the data for over 100,000 participants was released to the research community in 2021.

The presentation will focus on the scientific insights gained from the first release of the NMR metabolic biomarker data in UK biobank. The results reveal a prominent role of abundant circulating lipids and metabolites as risk markers beyond cardiometabolic diseases, including susceptibility to infectious diseases and risk for the onset of respiratory diseases, joint disorders and mental health outcomes. The talk will also highlight the application of the biomarkers for risk prediction of various common diseases to identify individuals at several fold increased risk compared to the remaining population. Release of the NMR biomarker data at scale in the UK Biobank highlights the promise of metabolic profiling in large cohorts for public health and translational research.

Proteomics in the era of large-scale biobanks
Professor Claudia Langenberg

Application of different proteomic technologies is now feasible at population scale. This talk will present examples of how the integration of proteomic with other omic technologies in large patient and population studies can help to predict disease risk, understand mechanisms, and reveal shared connections between diverse diseases.

Human cell atlas and its relevance to precision medicine
Muzlifah Haniffa

"Muzlifah has used functional genomics, comparative biology and single cell RNA sequencing to study the human immune system in health and disease. In this seminar, she will demonstrate the applications of single cell and spatial genomics to decode the developing human immune system.

SPEAKER BIOGRAPHIES

Professor Sir Rory Collins

Rory Collins is an epidemiologist who studies how to prevent and treat cardiovascular disease in large population-based studies. He trained in Medicine at St Thomas's Hospital, London University, and Statistics at George Washington and Oxford Universities. He has been at Oxford since 1981, where he is currently Head of the Nuffield Department of Population Health.

During the 1980s and early 1990s, Rory coordinated the ISIS "mega-trials" of the emergency treatment of heart attacks involving more than 130,00 patients. These trials showed that clot-dissolving and clot-preventing treatment could more than halve mortality, rapidly becoming part of routine care worldwide (as well as paving the way for selective use of non-pharmaceutical methods to open coronary arteries).

Since the early 1990s, he has been involved in conducting large-scale randomized trials of the effects of modifying blood levels of cholesterol. For example, the 20,000 patient Heart Protection Study that he led showed that lowering LDL-cholesterol with statin therapy safely reduces the risk of death and disability from cardiovascular disease among a much wider range of people than thought likely to benefit. As a consequence, statin therapy is now used extensively worldwide.

He became Principal Investigator of UK Biobank in 2005. Involving 500,000 participants, it is the largest deeply-characterized prospective study of disease globally, available for any type of health research. Over 30,000 researchers worldwide use it currently, generating 1700 papers in 2021 alone.

He was knighted for services to Science in 2011, elected to the Royal Society in 2015, and awarded the UK Medical Research Council's 2020 Millennium Medal for his national and international contributions to both cardiovascular disease and UK Biobank.

Richard Hebdon

I am the Director of Health & Life Sciences at Innovate UK, part of UK Research & Innovation (UKRI). I lead and manage the Innovate UK Health & Life Sciences Sector which comprises health, agriculture and food and biosciences, with a combined current portfolio valued at £685 million, and a historic investment of £1.9 billion.

My career spans 32 years and prior to joining Innovate UK, I worked in technology transfer, R&D management and research and innovation roles in the life science industry and public sector. This included working in medicines discovery and development, vaccine R&D and FMCG product innovation.

Professor David Hunter

David Hunter is the Richard Doll Professor of Epidemiology and Medicine and Director of the Translational Epidemiology Unit at the Nuffield Department of Population Health, University of Oxford, UK. He founded the Program in Genetic Epidemiology and Statistical Genetics at Harvard and was co-chair of the steering committee of the Breast and Prostate Cancer Cohort Consortium at the National Cancer Institute. He was co-director of the NCI Cancer Genetic Susceptibility Markers project focussed on genome-wide association studies, and Dean for Academic Affairs and Acting Dean at the Harvard TH Chan School of Public Health. He is the Chief Science Advisor to Our Future Health a major new national initiative in the UK that aims to return genomic information to consenting participants.

Dr Innocent Asiimwe

Innocent Gerald Asiimwe is a pharmacist who is originally from Uganda. He worked in Uganda and Botswana before pursuing further study in the UK at the University of Liverpool. Here, he obtained a MSc in Clinical Research, an MRes in Biomedical Sciences and Translational Medicine (Biology of Cancer), and a PhD in Pharmacology with a focus on warfarin pharmacogenomics in sub-Saharan Africa. He is currently working as a postdoctoral research associate in Prof Sir Munir Pirmohamed's group on an MRC-funded project evaluating the relationships between diseases, drugs and their therapeutic targets using existing large-scale data from electronic health records, genome-wide association studies and randomised clinical trials.

Dr Emma Magavern

Dr Magavern is a Clinical Pharmacology Registrar in London with an honorary affiliation with QMUL. She completed a BA in English prior to her MD and subsequent MScs in Bioethics and Genomics. Through training in clinical medicine, humanities, genetics and pharmacology she has developed an interest in the scientific merits, clinical potential and implementation challenges of pharmacogenomics. She was co-secretary of the RCP/BPS working group on pharmacogenomics.

Dr Daniel Martinez

Born in the eastern part of Spain, I studied Biology and got my PhD in Biomedicine at the Universidad de Valencia. My scientific interests lay in understand how microbes interact with each other and with the host. My PhD focused on the development of the theoretical framework to address the question of whether microbial dynamics can inform us about the health and disease state of the host. Under the supervision of a geneticist and a physicist, I was able to gain a new vision in biology at a systems level and develop the necessary bioinformatics skills to tackle the central questions of my PhD.

In 2018 I came to London to start a postdoc in Filipe Cabreiro's lab, where I could apply what I learned during my PhD and improve my experimental skills working with microbiology and C. elegans. The lab studies the metabolic interactions between microbes and host, which are essential if we want to understand the interplay between the two parts. During the pandemic I have polished my bioinformatics skills, developing several tools for the lab to study the microbial genetic diversity at a population scale and its relationship with host homeostasis in different drug contexts.

Jasmine Gratton

Jasmine completed her undergraduate degree in Anatomy and Cell Biology at McGill University. She then moved to London where she did an MRes in Biomedical Research at Imperial College London. Jasmine is now finishing her BHF-funded PhD programme in cardiovascular population genetics in Prof Aroon Hingorani's group at UCL.

Professor Henk-Jan Guchelaar PharmD PhD

Henk-Jan Guchelaar studied Pharmacy at the Rijksuniversiteit Groningen (RuG) and specialized as a hospital pharmacist and clinical pharmacologist. Since 2003, he is employed as a clinical pharmacist and clinical pharmacologist and professor of clinical pharmacy and chair of the department of Clinical Pharmacy & Toxicology at Leiden University Medical Center.

Since october 2008, he is also appointed professor of Clinical Pharmacy at the Faculty of Science, Leiden Academic Center for Drug Research, University Leiden and chair of the Leiden University focus area 'Translational Drug Discovery and Development'.
Pharmaceutical patientcare in oncology is his main area of clinical interest. He is program leader of the research program 'Personalised Therapeutics' investigating interindividual variability of drug response

with an emphasis on pharmacogenomics. He is (co-)author of more than 600 (Web of Science indexed) articles (Pubmed: 420) in international peer reviewed scientific journals. Guchelaar is coordinator of the EU funded Horizon 2020 project Ubiquitous Pharmacogenomics (www.upgx.eu) aimed at implementing pre-emptive pharmacogenomic testing in the EU.

From 2010-2016 he was a member of the national Central Committee on Research Involving Human Subjects, from 2003-2017, he was vice-chair of the Dutch Society for Clinical Pharmacology and Biopharmacy, and since 2016 he is member of the Dutch Medicines Evaluation Board. Since 2017, he is member of Council for Medical Sciences of the Royal Dutch Academy of Science and since 2020 member of the Academia Europea. Henk-Jan is founder of the new Master of Pharmacy, Leiden University Medical Center, University of Leiden.

Professor Douglas F Easton FMedSci FRS

Professor Douglas Easton is a leading genetic epidemiologist with a specific interest in the genetics of hormone related cancers. He graduated in mathematics from the University of Cambridge, and did his PhD in genetic epidemiology at the Institute of Cancer Research. Since 1995 he has been Director of the Centre for Cancer Genetic Epidemiology in Cambridge.
His studies have been instrumental in the characterisation of many cancer susceptibility genes, including BRCA1, BRCA2, CHEK2 and ATM, the identification of several hundred common cancer susceptibility variants, and the development of genetic risk scores. In 2007 he led the first genome-wide association study in breast cancer. Professor Easton leads the Breast Cancer Association Consortium, a collaboration involving more than 100 research groups and 300,000 subjects. He leads the EMBRACE cohort study of BRCA1/2 mutation carriers, and co-developed the BOADICEA risk prediction model, now used in genetic counselling worldwide.

Professor Matthew Brown

Matt Brown is a clinician-scientist who trained initially in medicine and rheumatology in Sydney, Australia before completing a Doctorate of Medicine based at University of Oxford, focusing on genetics of ankylosing spondylitis. He was appointed Professor of Musculoskeletal Sciences at University of Oxford in 2004. In 2005 Matt returned to Australia, firstly to University of Queensland, and since 2016, at Queensland University of Technology, where he was Professor and Director of Genomics. In 2013 he was elected to Fellowship of the Australian Academy of Sciences in recognition for his achievements in genetics research. In 2019 he moved to King's College London and Guy's and St Thomas' Hospitals NHS Trust to direct their NIHR Biomedical Research Centre, and in 2021 moved to the position of Chief Scientific Officer of Genomics England. He continues to work in genetics of human diseases, with a particular focus on common and rare bone and joint diseases, and in cancer genomics and personalized medicine. He continues to practice rheumatology, with a particular focus on spondyloarthritis.

Heli Julkunen

Heli Julkunen is a senior data scientist at Nightingale Health Plc, the Finnish innovator of an internationally recognized metabolomics platform for large cohorts, biobanks, and trials. She is co-leading Nightingale's data analyses on the UK Biobank, and has published scientific articles on biomarker discovery and risk prediction using the UK Biobank NMR biomarker data. She is a part of Nightingale's science team, leading various epidemiological analyses and focusing on translating the results from cohorts to consumer use.

Professor Claudia Langenberg

Claudia Langenberg is Professor of Computational Medicine at the Berlin Institute of Health at Charité (BIH) and MRC Investigator and Programme Leader at the MRC Epidemiology Unit at the University of Cambridge. Her research is focused on the genetic basis of metabolic control, and her team studies its effects on health through integration of molecular with clinical data in large-scale patient and population-based studies.
https://www.omicscience.org

Professor Muzlifah Haniffa

Muzlifah Haniffa is a Wellcome Senior Clinical Research Fellow, Professor of Dermatology and Immunology at Newcastle University and Associate Faculty, Cellular Genetics, Wellcome Sanger Institute.

She graduated from medical school in Cardiff, trained as a junior doctor in Cambridge and received her dermatology specialist training in Newcastle. Muzlifah is a Fellow of the Academy of Medical Sciences and a recipient of the Academy of Medical Sciences Foulkes Foundation Medal and the European Federation of Immunological Societies ACTERIA Prize in Immunology and Allergology.

Muzlifah is a leading member of the Human Cell Atlas initiative and pioneered the application of single cell genomics to decode the developing human immune system, and the human skin in health and disease

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genedrive plc is a molecular diagnostics company developing and commercialising an affordable, real-time PCR system that can serve as the basis for rapid detection of genetic variants, infectious diseases or pathogens aligned to clinical outcome.

The company most recently launched a ground breaking new pharmacogenetic test for use in emergency care, the Genedrive® MT-RNR1 ID Kit, a rapid point of care test to prevent antibiotic induced deafness in infants.

Manchester University Hospitals NHS Foundation trust is rolling out genedrive's point of care test that rapidly identifies babies carrying the MT-RNR1 m.1555A>G gene variant via a simple non-invasive cheek swab in just 26 minutes. For those carrying the variant, an alternative antibiotic can be prescribed, preserving hearing that would otherwise be lost on exposure to just a single dose of gentamicin.

The potential clinical usage of genetics at the point of care is boundless and will enable clinicians to create personalised treatment plans, ultimately providing patients the best possible chances for a positive outcome.

As we further develop our portfolio, we have an opportunity to reach millions of people with novel diagnostic tests. Ultimately, our goal is to positively change lives through rapid access to the benefits of molecular diagnostics. We are passionate about the opportunity to build a sustainable business around molecular diagnostics and to play an important role in the diagnostic and treatment challenges presented by global health issues.

Health in Code's core mission is the "identification of health problems that may benefit from a genetic diagnosis and the development and provision of genetic testing services for those conditions".
Its differential value lies in the combination of Knowledge Management tools and a dedicated expert multidisciplinary team, which allows it to attain a unique level of interpretation in their healthcare-focused genetic test reports. Pre- and post-test counselling offered by this select group of experts is what healthcare professionals value most.
Specialising in high-quality sequencing services (NGS) and interpretation of genetic test results, Health in Code offers physicians tools to bring personalized medical care to their patients. It is a European leader in the genetic diagnosis of inherited cardiovascular diseases and systematically deepens and extends the body of genetic information used for analysis to other areas, such as pharmacogenetics.
Fuelled by the desire for a more effective use of medications in healthcare settings, the pharmacogenomics area of Health in Code develop genomic products and services based on clinical evidence and utility.
Health in Code's approach is two-fold: a comprehensive one, with a global NGS pharmacogenetics panel that screens all genes and regions of interest included in the main clinical practice guidelines, and a targeted one, with panels including specific variants in genes such as DPYD, CYP2C9, and CYP2C19, among others, using different technologies, for example, qPCR, Sanger, SNaPShot, or MLPA.

The International Society of Pharmacovigilance (ISoP) is an international non-profit scientific organisation, which aims to foster Pharmacovigilance both scientifically and educationally, and enhance all aspects of the safe and proper use of medicines, in all countries.

MC Diagnostics Ltd. is an innovative molecular diagnostics company. We have designed, developed, and manufactured an automated low density array platform (ALDAS) which is currently providing proven clinical diagnostic assays in the fields of HLA typing for transplantation and the identification of specific HLA alleles implicated in predisposing patients to adverse drug reactions (ADRs).

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- Same day turnaround
- Fully automated detection

For more than a decade dnalife® has been at the forefront of genetic testing. Our laboratory reports aim to provide insights that can be used by the clinician to help ameliorate symptoms, reduce disease risk, and optimise their patients health. As one of the premier genetic testing laboratories, dnalife® offers an innovative approach to personalized medicine; offering a suite of nutrigenomic and pharmacogenomic tests that have been developed by our team of geneticists. We differentiate ourselves in the market through our extensive knowledge base, our selective test panels and our practical and user-friendly reports. We are committed to continually developing tests, and updating our reports, which truly help and guide practitioners and patients in optimizing health and create longevity through personalised healthcare.

Medcheck is a comprehensive pharmacogenomics test that analyses genetic variants associated with responsiveness to cardiovascular, psychiatric, pain and cancer medications. Prescribing medications according to an individual's genetic profile will improve drug efficacy, reduce drug-associated toxicity, and enhance patient outcomes.

Pacific Biosciences of California, Inc. (NASDAQ: PACB) is empowering life scientists with highly accurate sequencing platforms. The company's innovative instruments are based on Single Molecule, Real-Time (SMRT®) Sequencing technology, which delivers a comprehensive view of genomes, transcriptomes, and epigenomes, enabling access to the full spectrum of genetic variation in any organism. Cited in thousands of peer-reviewed publications, PacBio® sequencing systems are in use by scientists around the world to drive discovery in human biomedical research, plant and animal sciences, and microbiology. For more information, please visit www.pacb.com and follow @PacBio.

World Changing Science Deserves World Beating Support
Simplify your study operations with a full range of services from UK Biocentre
UK Biocentre's vast experience in sample services, hosting the NIHR Biosample Centre and as the largest UK COVID-19 "Lighthouse Lab" places us in the unique position to support, simplify, and streamline research, including.

- Cohort Studies.
- Diagnostics.
- Public Health initiatives.
- Precision Medicine.
- Genomics.
- Clinical Trails.

We can employ our knowledge and experience to help design, build and implement better, more efficient, cost-effective studies which allow our partners to focus on their science.
By seamlessly integrating our state-of-the-art robotic technology, quality-controlled logistics and ISO laboratory and storage workflows into our partner's study operations we enable our partners to

- Build and implement complete standards- compliant end-to-end studies.
- Identify and support the most complex study operations, whilst allowing them to maintain ISO quality standards.
- Design large scale studies supported by our unique high volume sample processing and storage services.

UK Biocentre is proud to be the partner of choice for some of the UK's largest and most prestigious, medical, and educational institutions.
They benefit from.

- Reduced costs and risks.
- Rapid study implementation.
- Flexibility.
- Total control.
- Quality-control

They recognise that study design and implementation does not need to be hard when they can rely on a partner with the experience, expertise, and capacity of UK Biocentre.

NEXT EVENTS

Open Targets workshop 18th October 2022

This workshop will bring together representatives from all sectors with an interest in Open Targets eco-system. It will discuss: -

- **Genomics in supporting therapeutic hypotheses**
- **AI in target prioritisation/therapeutic hypothesis generation**
- **The importance of phenotype - cellular, disease progression, patient stratification**
- **What questions do you ask to build a therapeutic hypothesis?**
- **How do you assess novelty of a target?**
- **How can pharmacogenetics help inform target prioritisation/therapeutic hypotheses?**

Decision Support workshop 9th November 2022

The Spine, Liverpool

This workshop will discuss: -

- **Needs of clinicians for decisions support tools to aid prescribing**
- **Standards framework for shared-decision-making support tools, including patient decision aids**
- **Statistical analysis for the development of decision support tools**
- **Developing a decision support tool to enable precision treatment**

If you are interested in joining either of these workshops please contact Christine cjmcn@liverpool.ac.uk

MEMBERSHIP

Membership is free please contact Christine cjmcn@liverpool.ac.uk

CPD CREDITS

8 CPD credits are awarded for attending the meeting, contact Christine cjmcn@liverpool.ac.uk for a certificate of attendance.

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