Genomic Diversity and Genomic Healthcare Conference 10.05.2022
Faculty

George P. Patrinos is Professor of Pharmacogenomics and Pharmaceutical Biotechnology in the University of Patras (Greece), Department of Pharmacy and holds adjunct Professorships at Erasmus MC, Faculty of Medicine, Rotterdam (the Netherlands) and the United Arab Emirates University, College of Medicine, Department of Pathology, Al-Ain (UAE). Also, he served for 12 years (2010-2022) as Full Member and Greece's National representative in the CHMP Pharmacogenomics Working Party of the European Medicines Agency (EMA), since 2018 Co-Chair of the Global Genomic Medicine Collaborative (G2MC) and since 2020 Editor-In-Chief of the prestigious Pharmacogenomics Journal, published by Nature Publishing Group.
George has more than 300 publications in peer-reviewed scientific journals, some of them in leading scientific journals, such as Nature Genetics, Nature Rev Genet, Nucleic Acids Res, Genes Dev and has co-edited the textbook "Molecular Diagnostics", published by Academic Press, now in its 3rd edition, and several other international textbooks, while he is the editor of "Translational and Applied Genomics" book series.
Apart from that, George is the main co-organizer of the Golden Helix Conferences, an international meeting series on Pharmacogenomics and Genomic Medicine.

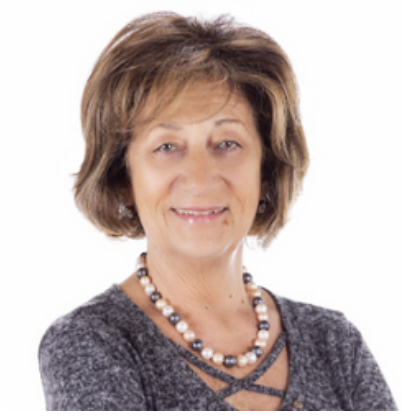
Professor Sir John Burn is a Professor of Clinical Genetics at the University of Newcastle, UK. Professor Sir John Burn obtained an MD with distinction, a first class honours degree in human genetics from Newcastle University, where he has been Professor of Clinical Genetics since 1991 and a consultant specialist since 1984. He led the regional NHS Genetics Service for 20 years and helped to create the Centre for Life which houses an education and science centre alongside the Institute of Genetic Medicine and Northgene Ltd, the identity testing company he launched in 1995. His research focuses on the prevention of cancer in those who have a high risk of the colo-rectal (bowel) cancer.

He chairs DNA device company QuantuMDx. He was knighted in 2010 and chosen as one of the first 20 'local heroes' to have a brass plaque on Newcastle Quayside in 2014. He received the Living North award in 2015 for services to the North East 2000 – 2015.

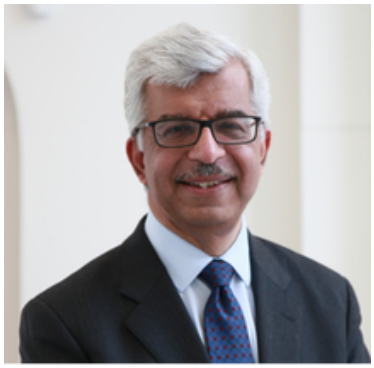
He is also Executive Chairman of the international organisation, the Human Variome Project, which seeks to share knowledge of genetic variation for clinical benefit. Since December 2017, Sir John has served as the Chairman of the Newcastle upon Tyne Hospitals NHS Foundation Trust.

Tuuli Lappalainen, PhD, is a Professor at KTH Royal Institute of Technology, the Director of the Genomics Platform and the National Genomics Infrastructure of SciLifeLab, Sweden, and an Associate Faculty Member at the New York Genome Center. Dr. Lappalainen received her PhD in Genetics from the University of Helsinki, followed by postdoctoral research at the University of Geneva and Stanford University. Her research focuses on functional genetic variation in human populations and its contribution to human traits and diseases, which her lab studies using both computational and experimental approaches. She has pioneered in integrating large-scale genome and transcriptome sequencing data to understand how genetic variation affects gene expression, which gives insight to biological mechanisms underlying genetic disease risk. She has contributed to many of the most important international research consortia in human genetics, including the 1000 Genomes Project, the Geuvadis Consortium, the GTEx Project, MoTrPAC, and TOPMed. She is a principal investigator of numerous NIH grants and a recipient of the Leena Peltonen Prize for excellence in human genetics, and the Harold and Golden Lamport Award in Excellence in Basic Research.

“Professor Damian Smedley leads a Computational Genomics team at Queen Mary University London where his research focusses on the use of phenotype data to obtain novel insights into disease causes and mechanisms. His team is involved in translational aspects for a number of projects such as the International Mouse Phenotyping Consortium (IMPC). In collaboration with other members of the Monarch Initiative he has developed tools that utilise phenotype comparisons for candidate gene prioritisation, particularly for whole genome sequence interpretation of rare disease patients as in the Exomiser software suite. Prof. Smedley served as Director of Genomic Interpretation at Genomics England from 2016-2018 and has led the analysis of the impact of the 100,000 Genomes Project pilot on rare disease diagnosis in healthcare.”



Dr. Angela R. Solano, Biochemist and Pharmacist
PhD in Medicine, University of Buenos Aires
Certified in Genetics (Argentinean Society of Genetics)
Chief Genotyping Laboratory, Department of Clinical Chemistry, University Hospital CEMIC
President, Argentinean Society of Genetics 2019-2021
Chair, Node of Argentina of the HVP/ HUGO
Co-Chair: Gene Disease/Specific Data Base Advisory Council, Education Committee and Scientific Advisory Committee, HUGO
Correspondent Member, "Committee on Molecular Diagnostics", International
Federation of Clinical Chemistry and Laboratory Medicine (IFCC)
Associated Editor, Human Genomics
Emeritus Member, American Society of Human Genetics.
Areas of interest: Genes Genotyping in germline and somatics in: cancer, endocrinology, cardiology, among the most frequent.



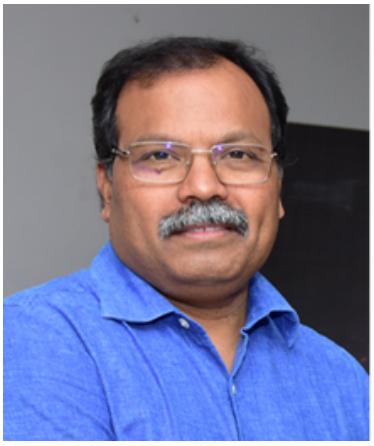
Professor Sir Munir Pirmohamed is the David Weatherall Chair of Medicine at the University of Liverpool, and a Consultant Physician at the Royal Liverpool University Hospital. He is also Director of the Centre for Drug Safety Sciences, and Director of the Wolfson Centre for Personalised Medicine. He is an inaugural NIHR Senior Investigator, Fellow of the Academy of Medical Sciences in the UK, Chair of the Commission on Human Medicines and is a non-executive director of NHS Improvement, and immediate Past President of British Pharmacological Society. He was awarded a Knights Bachelor in the Queen's Birthday Honours in 2015. His research is data-centred, and focuses on personalised medicine, genomics, clinical pharmacology and drug safety.

Thomas Borge, Blueprint Genetics is a PhD in Evolutionary Genetics from Uppsala University, Sweden. I have been working in several start-up companies within the field of genetics. For the last 6 years I have held the position as a Sales Director for Blueprint Genetics, a genetic testing company focused on inherited diseases. Blueprint Genetics was founded by clinicians in order to facilitate NGS diagnostics for clinicians. We are now having a global operation serving more than 4000 clinicians in 70 countries. Blueprint Genetics is since 2019 part of Quest Diagnostics, an American clinical laboratory and Fortune 500 company.

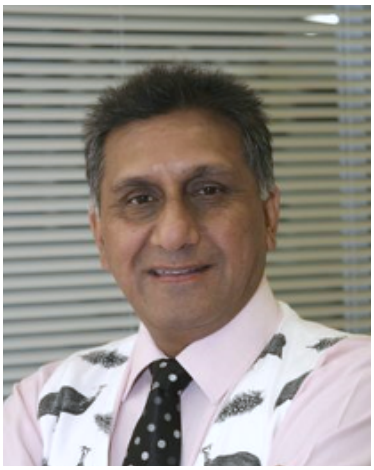
Teri A. Manolio, MD, PhD
Director, Division of Genomic Medicine, National Human Genome Research Institute (NHGRI)
Dr. Manolio is an internist and genetic epidemiologist who directs NHGRI's Division of Genomic Medicine, where she leads programs to develop and implement genomic applications in clinical care. She joined NHGRI in 2005 to lead efforts in applying genomic technologies to population research and clinical care, including the Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT) program, Undiagnosed Diseases Network (UDN), Clinical Sequencing Evidence Generating Research (CSER) Consortium, Electronic Medical Records and Genetics (eMERGE) Network, Implementing Genomics in Practice (IGNITE) Network, and the Clinical Genome (ClinGen) Resource. She continues to practice and teach internal medicine at the Walter Reed National Military Medical Center and is a professor of medicine at the Uniformed Services University of the Health Sciences. She is the author of over 300 research publications and has research interests in genomic risk prediction of complex diseases, ethnic differences in disease risk, integrating genomic research into electronic medical records, and incorporating genomic findings into clinical care.

Andreas Laner, Ph. D.
Head Genomics Program
MGZ – Medizinisch Genetisches Zentrum München
Personal Statement

I am a molecular biologist by training and have been active in cancer genetics and variant interpretation for more than 20 years; my research interests include hereditary cancer syndromes (especially Lynch/HNPCC syndrome, other hereditary colorectal cancer syndromes and HBOC). My position as Head of Genomics Program at MGZ includes supervision of the complete variant interpretation and variant curation process in our routine diagnostic lab as well as data sharing, curation and knowledge transfer initiatives.



Kumarasamy Thangaraj is the Director of Centre for DNA Fingerprinting and Diagnostics at Hyderabad. Prior to this, he was Chief Scientist and Group Leader at the Centre for Cellular and Molecular Biology (CCMB), Hyderabad. His main research interests are; origin of modern human, cardiovascular diseases, mitochondrial disorders, and male infertility. He has published over 275 research articles, including Cell, Lancet, Science, Nature, Nature Genetics, etc. He is an elected Fellow of - Indian Academy of Sciences, Indian National Science Academy, and National Academy of Sciences. He is a recipient of several awards, including J C Bose Fellowship, Sun Pharma Research Award in Medical Sciences, Raman Research Fellowship, Life-time achievement Award, Excellence in Science Award, Distinguished Scientist Award, etc. He is a Board Editor of Mitochondrion; Associate Editor of BMC Medical Genetics; BMC Genetics; Tropical Medicine and International Health, and member of the Editorial Board of the journals – Scientific Reports, Human Genetics, and Clinical Genetics. He was the President of the Indian Society of Human Genetics (2011 – 2015) and the founder of the Society for Mitochondrial Research and Medicine.



Raj Ramesar is Professor and Head of the Division of Human Genetics, as well as Head of the Department of Pathology, at the University of Cape Town and its Affiliated Hospitals in South Africa. This facility has wide-ranging clinical responsibilities from quaternary and tertiary care levels, to extensive rural outreach programmes, in addition to diagnostic and research capabilities. His interest is in using the exciting developments in the field of genomic sciences to investigate human biodiversity. Africa offers the opportunity to use population lineages in all of their richness towards identifying aspects of human biology, that have to do with both health and disease.

As the Director of the MRC Genomic and Precision Medicine Research Unit, the emphasis of his research has been on disease susceptibility in South African populations, progressing from the commonly-recognised inherited diseases, to those that are more complex yet more common and relevant to a large burden of disease. In this regard, his main research foci are on Hereditary Cancers, Inherited Retinal Degenerative Diseases, and Psychiatric Genomics. Raj recently received the (Vice Chancellor's) Alan Pifer Award for 'outstanding research in cancer genetics which shows relevance to the advancement of South Africa's disadvantaged populations'. He was also elected to the College of Fellows of the University of Cape Town. Apart from being on the editorial board of several international journals, Raj serves as co–chair of the Scientific and Medical Advisory Board of Retina South Africa, he is on the Executive of the African Society for Human Genetics, and is the latter organisation's Liaison Officer to the International Federation of Human Genetics Societies. As Chair of the Local Organising Committee, he organised the Joint Conference of the African and Southern African Societies in Cape Town. Raj also led a successful bid to host the International