Global Genomic Medicine Collaborative (G2MC)

6th International Conference

September 29 - October 1, 2021

Speaker & Moderator Booklet

G2MC 6th International Conference Speaker List

Marc Abramowicz, MD, PhD

Switzerland
Professor and Head, University of Geneva

Marc Abramowicz, MD, PhD, is a clinical geneticist, Professor at the University of Geneva, Switzerland, and Head of the Center of Genomic Medicine of the Geneva University Hospitals, an interdisciplinary platform currently aiming at identifying the molecular causes of unidentified disease and precision medicine. He is also a Full Professor at the Université Libre de Bruxelles, Belgium. Dr Abramowicz has authored more than 100 research papers relating to genetics or genomics in peer-reviewed journals (GoogleScholar h-index 47).

Bassam Ali, PhD

United Arab Emirates
Professor, United Arab Emirates University

Dr. Bassam R. Ali is a professor in Molecular and Genetic Medicine at the College of Medicine and Health Sciences, United Arab Emirates University. Prof. Ali obtained his PhD degree in biochemistry from the University of Cambridge in 1994 then worked at Imperial College London and University College London before joining the United Arab Emirates University in August 2006. His current research interests are: (1) Elucidation of the cellular and molecular mechanisms underlying human genetic diseases (2) The identification of disease genes responsible for rare recessive disorders in Arab populations and (3) Pharmacogenomics. Prof. Ali published over 130 original articles and reviews.

Fahd Al-Mulla, MD, PhD, FRCP

Kuwait

Chief Scientific Officer, Dasman Diabetes Institute

Fahd Al-Mulla is the Chief Scientific Officer and Heads the Department of Genetics and Bioinformatics at Dasman Diabetes Institute, a Professor of Molecular Pathology and Genomic medicine at Kuwait University. Prof Al-Mulla received his medical License and PhD from Glasgow University. He is a Fellow of the Royal College of Physicians Of Edinburgh. Prof Al-Mulla's Research activities focus on cancer and Diabetes, he has pioneered the use of next generation sequencing and microarrays in diagnostics and Precision Medicine. He has extensive experience in Genomic related technologies and intellectual property development. He currently holds 6 patents related to Diabetes, wound healing and cancer. Prof Al-Mulla serves as a chair of the Evidence group in the Global Genomic Medicine Collaborative and of the International Confederation of Countries Advisory Council of the Human Variome.

In 1993, Prof Al-Mulla graduated in Medicine from Glasgow University. He graduated with a PhD in 1999, studying the molecular basis of colorectal cancer metastasis supervised by the late Professor George Burnie.

Prof Al-Mulla established 2 laboratories at Kuwait University, a large research core facility that serves about 600 academics, students and researchers and the latter is a molecular diagnostic laboratory that he heads. His research led to the identification of two novel metastasis suppressor genes, namely Carbonyl Reductase and Raf Kinase inhibitory Proteins, which he and his team characterized further and their therapeutic roles in cancer patented. In recent years Prof Al-Mulla's research focused more intensely on wound healing and angiogenesis defects in a genetic rat model of type-2 diabetes mellitus. He and his colleagues were not only able to ameliorate insulin resistance in these rat models, but also precisely dissect the the precise molecular defects predisposing the animals to diabetes. Prof Al-Mulla spearheads an ambitious project to whole genome sequence thousands odd individuals from Kuwait. He currently established a database of individuals who were whole genome or exome sequenced with variety of aliments including cancer, multiple sclerosis and normal individuals. These data are of exceptional value for understanding population specific variants and distinguishing them from pathogenic variants.

Prof Al-Mulla serves as a chair of the Evidence group in the Global Genomic Medicine Collaborative and served as a chair of the international confederation of countries Advisory Council of the Human Variome. His ResearchGate and Orchid scores are higher than 97.5% of researchers world wide. Prof Al-Mulla is a highly cited researcher. He is also a keen blogger, Nature Biotechnology blog- The Bioentrepreneur blog, Trade Secrets, boasts contributors from around the globe, providing regular insight and commentary from those helping build the world's biotechnology sector. He also attained six patents over the course of his career thus far, with more anticipated in the years to come.

Alba Ancochea

Spain

Director, FEDER y Fundacion FEDER

Alba Ancochea, FEDER director since 2013 and leading its Foundation since 2015.

With a degree in Psychology and a degree in Special Needs Teaching, holding a master's degree in Human Resources Management and Non-profit Organization Management (NGO), Alba currently leads a team of almost 50 people who work advocating in favour of people living with rare diseases (PLWRD) and without a diagnosis, favouring their inclusion and generating strategies that contribute to improving their quality of life.

Her career in the Federation started in 2009 as part of the Psychological Attention Service (SAP in Spanish). Through her experience, she had the opportunity to know in depth the reality of this community, an environment that was already familiar to her due to her personal experience with these pathologies.

Since then, Ancochea has always been an example of change and transformation, helping to qualify the representation of patient organizations in decision-making bodies that affect them. Thanks to her experience and training, she has taken part in the development and monitoring of the main European roadmaps like the 'State of the art' project; at the national level with the Strategy for Rare Diseases of the National Health System of Spain; as well as of the autonomic plans related to each region of the country.

This action has been complemented with the representation of patients in related bodies, such as Comité Científico Asesor del Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER) - Scientific Advisory Committee of the Centre for Biomedical Research in Network of Rare Diseases-, or other key platforms.

At the international level, Alba Ancochea is part of the Rare Diseases International (RDI) Council, promoting the involvement of Spain in events that have marked a milestone for rare diseases, such as the event that the RDI and the NGO Committee for Rare Diseases held at the United Nations headquarters in 2019.

Likewise, she has been re-elected for the second time as a member of the Board of Directors of Rare Diseases Europe – EURORDIS-. Thanks to this, projects in the European health field have been rolled out with greater depth in Spain, such as the European Reference Networks, which seek to improve knowledge on rare diseases or initiatives of a social nature, such as the INNOVCare Project, which aims to guarantee holistic care of PLWRD.

Additionally, she represents FEDER at the EURORDIS Council of National Alliances (CNA), the governing body of the European Network of National Alliances, which is made up of European National Alliances, recognized as such by the EURORDIS BoD. Through collaboration at the European level and networking through EURORDIS, national rare disease alliances share information, experiences and good practices.

Ancochea also works as Advocacy Advisor at ALIBER (the Ibero-American Alliance for Rare Diseases) since its creation in 2014. This organisation is as of today a network that represents over 500 organizations of patients with rare diseases in 17 countries.

The representation of the whole of the Ibero-American movement in these international bodies has been completed with her incorporation to the International Rare Diseases Research Consortium (IRDiRC) Patient Advocacy Constituent Committee (PACC). As part of it, Ancochea represents the entire Ibero-American Alliance for Rare Diseases, intending to integrate the perspective of patients in the largest research body for these pathologies worldwide.

Rodrigo Andaur, PhD

Chile

Biochemist, Comision Chilena de Energia Nuclear

My name is Rodrigo Andaur, I'm from Chile, a small country located south of the world. I Started my scientific life studying biochemistry and then made my doctoral studies in radiobiology. Currently, I'm making my postdoctoral fellowship, working in cell-cell communication in radiation context. As pandemic consequence, I started a new investigation, using several database, in order to understand the molecular deregulation during cancer progression.

Laura Arbour, MD

Canada

Professor, University of British Columbia

Dr. Laura Arbour, is a Professor in the Department of Medical Genetics at the University of British Columbia, situated at the Island Medical Program on Vancouver Island. Trained as both pediatrician and clinical geneticist (McGill University), her multifaceted Community Genetics Research Program, addresses rare, single gene disorders and more complex genetic conditions affecting Indigenous patients. Her clinical practice is focused on diagnosis of rare genetic disease and cardiogenetics. She is currently the project lead on a Genome Canada/Genome BC/CIHR funded Large Scale Applied Research Project, entitled “Silent Genomes: Reducing health-care disparities and improving diagnostic success for Indigenous children with genetic disease” which aims to address the challenges of inequitable access to genetic/genomic diagnosis and care for Canadian Indigenous populations.

Ricardo Armisén, MD, PhD

Chile
Professor, Universidad del Desarrollo

Ricardo Armisén (M.D., Ph.D.) is a Professor and Researcher at the Center of Genetics and Genomics at Facultad de Medicina Clinica Alemana Universidad del Desarrollo in Santiago, Chile.

Before his current position, Dr. Armisén had the privilege to lead two Comprehensive Cancer Research Centers (one at Universidad de Chile and one at Pfizer) in Chile and be part of major international collaborative efforts, with the support of the National Cancer Institute (USA), Pfizer, Thermo Fisher Scientifics, Chilean Grant agencies (CORFO and CONICYT) and the Universidad de Chile, to build, from scratch, cancer genomics, biobanking and molecular diagnostics facilities and laboratories to develop cancer genomics research in South America.

Dr. Armisén earned a Ph.D. in Biomedical Sciences from U. de Chile and a Medical degree from the same institution.

Esther Rodriguez Blanco, PhD

Spain
Head of the European Project Office, ISCII

With a PhD in Physics and a postgraduate degree in International Management of R&I projects, Esther Rodriguez has dedicated her professional life to R&I management and international cooperation. Appointed as head of the European Project Office of ISCIII (National Institute for Health in Spain), she has had the opportunity to participate in different international projects and initiatives related to Personalized Medicine. She coordinates the EULAC PerMed consortium, devoted to build bridges between the LAC region and Europe in PM. She is also part of the European initiative “1 Million Genome” and EU-AFRICA PerMed project.
She is also an active member of the Horizon Europe NCP network, with expertise in Health, Research Infrastructures and ERC.

Nadia Carstens, PhD

South Africa

Principal Medical Scientist, University of the Witwatersrand

Dr Carstens is a medical scientist specializing in the field of human genetics. She joined the Division of Human Genetics (National Health Laboratory Service and University of the Witwatersrand) as a principal medical scientist in 2016. She has a particular interest in the application of next-generation sequencing technology to improve diagnostics for rare Mendelian disorders in African populations. She is an active member and past chair of the H3Africa Rare Diseases Working Group and co-investigator on the Deciphering developmental disorders in Africa (DDD-Africa) project.

Vajira Dissanayake, MBBD, PhD, FNASSL, FIAHSI

Sri Lanka

Head & Chair Professor of Anatomy; Faculty of Medicine, University of Colombo, Sri Lanka

Professor Vajira H. W. Dissanayake MBBS (Colombo), PhD (Nottingham), FNASSL, FIAHSI is the Chair and Senior Professor in the Department of Anatomy, Genetics, and Biomedical Informatics and the Dean of the Faculty of Medicine, University of Colombo. He is a pioneer in Genetics, Genomics, Biomedical Informatics and Bioethics in Sri Lanka. He is a leading researcher in these fields in Sri Lanka. He has supervised 15 PhD students and 60 MSc students. He together with his students and colleagues have been the recipient of more than 40 awards for research and innovation at university, national and international levels. In recognition of his scientific achievements he was elected a fellow of the National Academy of Sciences of Sri Lanka in 2013, a fellow of the International Academy of Health Sciences Informatics in 2020, and conferred the national titular honour of vidya jyothi in 2019. In 2014 he was invited to a meeting of the Global Leaders in Genomic Medicine in Washington DC, USA by the Institute of Medicine, USA and appointed to the founding board of the Global Genomic Medicine Collaborative (G2MC) that was established following that meeting. The G2MC is now championing the implementation of genomic medicine in emerging countries around the world.

Prof. Dissanayake has held many leadership positions in the field of medicine. He was the President of the Sri Lanka Medical Association in 2012 and the President of the Commonwealth Medical Association from 2016 to 2019. The other leadership positions held by Prof. Dissanayake include Honorary Secretary, Sri Lanka Medical Association (2000 and 2005); Founder Secretary, Health Informatics Society of Sri Lanka (1998-2000); President, Health Informatics Society of Sri Lanka (2009-2019); President, Asia Pacific Association for Medical Informatics (2019-2020); Vice President (Asia Pacific), International Medical Informatics Association (2019-2020); Board Member, Steering Committee, Forum for Ethical Review Committees in Asia and the Western Pacific (since 2010); Chairperson, Commonwealth Centre for Digital Health (since 2018); and Chairperson, Commonwealth Health Professions and Partners Alliance (since 2021).

Antu Kumar Dutta, MD

India

Assistant Professor, Department of Biochemistry, AIIMS Kalyani

Professional Achievements:

Junior Resident: PGIMER, Chandigarh [2008-2011]

Senior Resident : Clinical genetics Unit, Christian Medical College, Vellore [2013-2015]

Clinical Molecular Geneticist : NIBMG Kalyani [2015-2019]

Assistant Professor : IPGMER & SSKM Hospital, Kolkata[2011 - 2013]

Publications:

32 publications in National and International journals with more than 350 citations.

Research interests:

Population Genetics, Molecular & Biochemical Genetics

Email Id: atanu.biochem@aiimskalyani.edu.in

Kelly East, MS, CGC

USA

Lead Genetic Counselor, HudsonAlpha Institute for Biotechnology

Kelly leads the provision of genetic and genomic counseling for research projects and clinical services at HudsonAlpha, as well as the development of educational experiences and resources for healthcare providers, trainees, and patients.

Bogi Eliasen, MA

Denmark

Director of Health, Copenhagen Institute for Futures Studies

Clara Gaff

Australia

Executive Director, Melbourne Genomics Health Alliance

Clara Gaff is Executive Director of Melbourne Genomics Health Alliance. Clara sits on the Australian Genomics' National Steering and National Implementation Committees and co-led their Education and Workforce Development program.

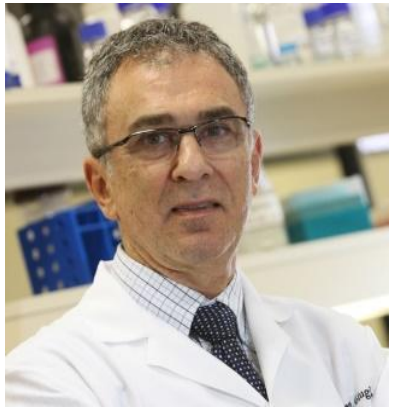
Clara has been involved in the use of genetics and genomics in healthcare through roles in genetic counselling, management, health professional education, and strategic development in Australia and the UK. She has worked in public health, government, academic and not-for-profit sectors. Her leadership has been recognised through awards from the National Society of Genetic Counselors (USA) and BioMelbourne Network.

Geoff Ginsburg, MD, PhD

USA

Director, Duke Center for Applied Genomics and Precision Medicine & Co-Chair and President, Global Genomic Medicine Collaborative

Dr. Ginsburg is the founding director of the Center for Applied Genomics & Precision Medicine at the Duke University Medical Center and of MEDx, a partnership between the Schools of Medicine and Engineering to spark and translate innovation. His research addresses the challenges for translating genomic information into medical practice and the integration of precision medicine into healthcare.



Roberto Giugliani, MD, PhD, MSc

Brazil

Professor, Department of Genetics, Federal University of Rio Grande do Sul (UFRGS)

Dr. Roberto Giugliani, MD, PhD, Full Professor at the Department of Genetics of the Federal University of Rio Grande do Sul, is a medical geneticist who founded and is an active member of the Medical Genetics Service of the University Hospital, in Porto Alegre, Brazil. He is member of many international committees and consultant for the DASA/GENEONE group, and other companies. He is also Editor-in-Chief of the Journal of Inborn Errors of Metabolism and Screening, Chairman of the Latin American School of Human and Medical Genetics, and Member of Brazilian Academy of Sciences. He is past President of the Latin American Society of Inborn Errors of Metabolism and Newborn Screening, the Latin American Network of Human Genetics, and the Brazilian Society of Medical Genetics and Genomics, and former Director of the WHO Collaborating Centre for the Development of Medical Genetics Services in Latin America. Prof. Giugliani's main interests are concentrated in screening, diagnosis, and treatment of inborn errors of metabolism, particularly of lysosomal storage diseases, having supervised the training of over 100 MSc and PhDs, and being author of more than 500 scientific papers.

Carolina Goic

Chile

Senator of the Republic of Chile

Carolina Goic Boroevic was born on 20 December 1972 in Santiago, Chile. Since 2013 she has been senator of the Republic of Chile for the Region of Magallanes and Chilean Antarctic. She is a Social Assistant and Economist who graduated from the Pontificia Universidad Católica de Chile.

In her parliamentary work she places emphasis on: initiatives that seek to reconcile work and family life; equality of rights and remuneration between men and women; protection of motherhood; prevention of alcohol and drug use in the youth; the integral improvement of health with emphasis on the promotion of a national cancer law; the mental health of the population; and care of the environment.

Claudia Gonzaga-Jauregui, PhD

Mexico

Principal Investigator, International Laboratory for Human Genome Research, LIIGH-UNAM

Claudia Gonzaga-Jauregui grew up in Mexico where she did her undergraduate studies in Genomic Sciences at the National Autonomous University of Mexico (UNAM). She obtained her PhD in Molecular and Human Genetics from Baylor College of Medicine, where she contributed to large population genomic studies such as HapMap 3 and pioneered the analyses of genomic sequencing data for the identification of disease genes and molecular diagnoses. Since then, her research has focused on the investigation of human pathogenic and polymorphic genomic variation that contribute to human traits and diseases. Her research focuses on family-based analyses of rare and common genetic disorders to better understand disease mechanisms and pathophysiology. Throughout her career, she has led large-scale Mendelian genomics projects in academia and industry to identify medically relevant variation and identify novel disease associated genes. Claudia has now established her research group focused on Mendelian genomics and precision health at the International Laboratory for Human Genome Research (Laboratorio Internacional de Investigación sobre el Genoma Humano, LIIGH) of UNAM in Mexico. She believes that the application and understanding of human genetics and genomics can lead to improved treatments and the realization of precision genomic medicine for everyone around the globe.

Evelin González, BS

Chile

Bioinformatics Engineer, Universidad del Desarrollo

My name is Evelin González, I am a Bioinformatics Engineer from the University of Talca. I finished my studies in 2015. For the last three years, I have developed bioinformatics projects in biomedicine. I am a research assistant at Functional Cancer Genomics Laboratory, Facultad de Medicina, Universidad del Desarrollo, Chile. I am working on the Pan-cancer project "Discovery of New Genomic Alterations in Cancer Patients from South America," analyzing more than 2,000 tumor samples to identify new and known mutations in driver genes. The objective is to identify new processable mutations in known cancer driver genes present in NGS Assay and build a database of genomic variation for cancer genes in Chile, Peru, and Brazil.

Lovemore Gwanzura, B.sc. Mphil medicine, MMsc.clin epi, PHD.

Zimbabwe
Professor, University of Zimbabwe

Prof. Lovemore Gwanzura is a clinical Microbiologist and epidemiologist. He is professor in the Faculty of Medicine and Health sciences. He is a senior Lecturer in the Department of Diagnostics and investigative sciences. Where lecturers in microbiology and molecular biology and genetics as well as carry out research work currently with 115 peer reviewed scientific papers.

Pratiksha Gyawali, MBBS, MD

Nepal
Consultant Biochemist, Dhulikhel Hospital & Lecturer of Clinical Biochemistry, Kathmandu University, School of Medical Sciences

Pratiksha Gyawali MBBS, MD (Nepal) is a consultant biochemist at Dhulikhel Hospital and a Lecturer of Clinical Biochemistry at Kathmandu University, School of Medical Sciences, Nepal. She received her MBBS from KUSMS, and MD from Institute of Medicine, Tribhuvan University of Nepal under merit postgraduate scholarship.

Pratiksha has been a pioneer in her country in promoting FHH being engaged in a series of activities with colleagues or serving the general public. She aspires to develop clinical research skills and foster genomic medicine in terms of patient care, diagnostic services and education in Nepal. To name a few, she has more than five years of experience in educating students of diverse background of medical and allied health sciences. Pratiksha is also an active member of Nepalese Association of Clinical Chemistry (NACC) and has a great contribution as an organizing member of national level workshops to uplift the quality assurance in the clinical laboratories of Nepal by collaborating with American Association of Clinical Chemistry (AACC), International Federation of Clinical Chemistry (IFCC) and University Grant Commission (UGC) Nepal. As a project committee member, she has provided her expertise in the

ongoing project entitled “Nurses led continuum of care approach for addressing diabetes in Nepal” under World Diabetes Foundation and “Community based life style intervention for diabetes management in Nepal (GACD-AMED).

In 2019, Pratiksha participated in the “International Summit In human genetics and genomics” at NHGRI/ NIH and later attended “Implementation Science School Training” organized by GACD. Most recently, she is corresponding member from Nepal in the IFCC Task force in “Global Newborn Screening” and Young Investigator and member of Family Health History Flagship project at Global Genomic Medicine Collaborative supporting its mission and efforts.

Christian Happi, PhD

Nigeria

Director, African Centre of Excellence for Genomics of Infectious Diseases, REDEEMER'S UNIVERSITY, NIGERIA

Christian Happi, is a Professor of Molecular Biology and Genomics and Director of the World Bank funded African Center of Excellence for Genomics of infectious Diseases (ACEGID) in Redeemer’s University, Ede, Osun State, Nigeria.

Professor Christian Happi, did his postdoctoral fellowship at Harvard University from 2000-2003. He subsequently worked at Harvard University as a Research Scientist (2004-2007) and became an adjunct Professor at Harvard University School of Public Health between 2007-2011.

Professor Happi in an unprecedented way, recently used next generation sequencing technology to perform the first sequence of the new SARS-CoV-2 in Africa, within 48 hours of receiving sample of the first case in Nigeria. This seminal work not only provided an insight into the detailed genetic map of the new coronavirus in Africa, not only confirm the origin of the virus, but also pave the way to the development of new countermeasures including new diagnostics, therapeutics and vaccines.

He received the Merle A. Sande Health Leadership Award in 2011; the 2016 Award of Excellence in Research, by the Committee of Vice- Chancellors of Nigerian Universities; the 2019 Human Genome

Organization (HUGO) Africa Prize for his seminal work on infectious diseases genomics in Africa, including Ebola and Lassa fever and the 2020 Bailey K. Ashford Medal by the American Society of Tropical Medicine and Hygiene (ASTMH).

Rich Haspel, MD, PhD

USA

Associate Professor, Beth Israel Deaconess Medical Center and Harvard Medical School

Dr. Haspel is currently the Vice Chair for Education in the Beth Israel Deaconess Medical Center (BIDMC) Department of Pathology and an Associate Professor of Pathology at Harvard Medical School. Since 2012, he has received over $2 million in grant funding from the United States National Institutes of Health to facilitate the work of national committees in developing genomics curricula for pathology residents and medical students. He is also Co-Chair of the National Human Genome Research Institute's Inter-Society Coordinating Committee for Practitioner Education in Genomics. (ISCC-PEG) which aims to improve genomic literacy of healthcare providers and enhance the effective practice of clinical genomic medicine by facilitating interactions among the key stakeholders in genomics education.

Professor Dame Sue Hill

United Kingdom

Chief Scientific Officer and Senior Responsible Officer for Genomics in NHS England and NHS Improvement

Professor Dame Sue Hill is the Chief Scientific Officer for England. Sue works across the health and care system as the head of profession for the healthcare science workforce in the NHS and associated bodies who work in over 50 different scientific specialties in the NHS in England. In addition, Sue is the Senior Responsible Officer for Genomics in NHS England and is responsible for overseeing the implementation of the NHS Genomic Medicine Service.

Norita Hussein, MBBS, MFamMed, PhD

Malaysia

Primary Care Physician, Faculty of Medicine, Universiti Malaya

Dr. Hussein is an Academic Lecturer and Primary Care Physician in the Department of Primary Care Medicine, Faculty of Medicine, Universiti Malaya. As a primary care physician, she has been responsible in the provision of definitive care to patients at primary care level, ensure continuity of care and comprehensiveness of patients' wellbeing especially with chronic medical illnesses. She is greatly involved in development of effective strategy to improve the primary care practice. As one who encounters the public, one of her great passion is to improve the way primary care providers communicate and manage individuals about genetic risk. Her research work in Malaysia has involved exploring primary care providers' management of carrier screening as well as exploring the views and experiences of carriers about thalassaemia screening. She is particularly interested in the special population; indigenous population and adolescent where decision making about inherited conditions and reproductive issues could be influenced by social, religious, cultural beliefs and level of health literacy.

Desalyn Johnson

USA

Medical Student, University of Alabama at Birmingham

Desalyn Johnson is a medical student at the University of Alabama at Birmingham. She graduated summa cum laude from the University of Alabama in Huntsville with a Bachelor of Science in Biological Sciences and a minor in Spanish Language. She investigated the effects of essential oils on MCF-7 breast cancer cells. Her manuscript, "The Cytotoxic Effects of Essential Oils on Estrogen Receptor Positive MCF-7 Breast Cancer Cell Line", was published in the student peer reviewed journal Perpetua. Her paper, "The first post-natal clinical description of true mosaic complete tetrasomy 21: A case report", was recently published in the American Journal of Medical Genetics. Under the tutelage of Drs. Bruce Korf and Richard Haspel, she analyzed data from the G2MC Educational Needs Assessment Survey to determine best ways to internationally implement genetic/genomic educational programs. She has presented the findings to the G2MC Education Working Group and the manuscript is presently under review. Johnson is currently pursuing a National Institutes of Health (NIH) funded National

Research Service Award to investigate the effect of insurance status on infant mortality and morbidity rates in the Unites States. She is interested in pediatrics, health care reform, and ameliorating health care disparities. Her other interests include fitness, language learning, and literature.

Julie Johnson, PharmD

USA

Dean and Distinguished Professor, University of Florida, College of Pharmacy

Julie A. Johnson, Pharm.D., is Dean of the University of Florida College of Pharmacy and Distinguished Professor of Pharmacy and Medicine. She is an international expert in cardiovascular pharmacogenomics and genomic medicine implementation, for which she has received nearly $50M in research funding and been named a Clarivate Analytics Highly Cited Scientist in 2015, 2016, 2017 and 2018, indicating she was in the top 1% of the most highly cited scientists in the her field globally in the decade preceding each year on the list. Dr. Johnson has served in numerous capacities with the NIH, the Food and Drug Administration, and leadership roles in multiple professional societies, including as President of the American Society of Clinical Pharmacology and Therapeutics. She has received numerous awards and honors and was elected to the National Academy of Medicine in 2014.

Thillainathan Kobika

Sri Lanka

Lecturer (Prob), University of Jaffna

Ms. Kobika Thillainathan is a lecturer and researcher at the Faculty of Allied Health Sciences, University of Jaffna. Kobika received the B.Sc. (Hons) in Medical Laboratory Sciences, from the University of Sri Jayewardenepura, Sri Lanka (2015), and M.Sc. degree in Molecular Pathology (reading) at the University of Colombo, Sri Lanka. She possesses the experience of working as a scientific officer who worked majorly on DNA typing with a scientific approach. Her research interests include genetics behind complicated human diseases especially, the studies related to cardiovascular genetics. She teaches courses in undergraduate degree and has a hand in supporting undergraduates in research

and publication since 2018. She is highly interested in interdisciplinary research and having co-authored a book on 'Palmyrah Research in Sri Lanka: A Way Forward' which was published in 2019. She has been an active member of Research and Publication Committee of the Faculty of Allied Health Sciences, University of Jaffna and currently is involved in creating the first molecular research laboratory within the faculty to serve as an initiative in the Northern region.

Bruce Korf, MD, PhD

USA

Associate Dean for Genomic Medicine, The University of Alabama at Birmingham

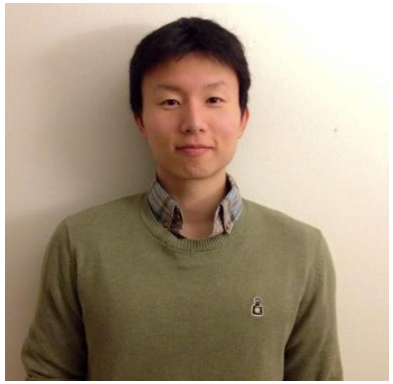
Dr. Korf is the Associate Dean for Genomic Medicine, School of Medicine; Chief Genomics Officer, UAB Medicine; Wayne H. and Sara Crews Finley Endowed Chair in Medical Genetics, Professor of Genetics, Co-Director of the UAB-HudsonAlpha Center for Genomic Medicine, Associate Director for Rare Diseases, Hugh Kaul Personalized Medicine Institute and editor-in-chief of the American Journal of Human Genetics. Dr. Korf is past president of the Association of Professors of Human and Medical Genetics, past president of the American College of Medical Genetics and Genomics, and current president of the ACMG Foundation for Genetic and Genomic Medicine. He has served on the Board of Scientific Counselors of the National Cancer Institute and the National Human Genome Research Institute at the NIH. His major research interests are genomic medicine and the natural history, genetics, and treatment of neurofibromatosis. He serves as principal investigator of the Department of Defense funded Neurofibromatosis Clinical Trials Consortium, and co-PI of the Alabama Genomic Health Initiative and the All of Us Southern Network. He is co-author of Human Genetics and Genomics (medical student textbook, now in fourth edition), and Emery and Rimoin's Principles and Practice of Medical Genetics.

Stefania Koutsilieri, MSc.

Greece
PhD Student, Karolinska Institute, Sweden

Stefania Koutsilieri is a Ph.D. candidate in the Laboratory of 'Personalized Medicine and Drug Development' in the Physiology and Pharmacology Department of Karolinska Institutet, under the supervision of Assoc. Prof. V.M.Lauschke. After graduating with honors from the Pharmacy School of the National and Kapodistrian University of Athens (NKUA), she completed her Master's degree in Pharmacology. Stefania has been awarded a Ph.D. scholarship from the Onassis Foundation to pursue research in the fields of Pharmacology and Pharmacogenomics.

Her research focuses on the profound characterisation of the signaling pathways activated upon the binding of medications on G protein-coupled receptors (GPCRs) in an effort to gain a better understanding of drug efficacy and side effects. The impact of interpatient variability is also investigated by employing 3D culture models of primary human tissues. Apart from the research projects in which she is involved, Stefania deeply believes that for Pharmacogenomics to be implemented in the clinic, it is of tremendous importance that regulatory aspects should be taken into consideration as carefully as research itself. To this end, Stefania focuses on the delineation of the similarities and most importantly, the discrepancies between the pharmacogenomic information that is documented in the drug labels approved by the two major regulatory bodies (FDA, EMA) and renowned research consortia, like the Clinical Pharmacogenetics Implementation Consortium (CPIC; https://cpicpgx.org).



Seung-been "Steven" Lee, PhD

South Korea
Senior Researcher, Macrogen Inc.

Dr. Seung-been "Steven" Lee received his BS in Biochemistry and PhD in Genome Sciences from the University of Washington, Seattle, WA. He is currently a senior researcher at Precision Medicine Institute of Macrogen Inc. where he has been leading various R&D projects as a bioinformatician, focusing on the fields of pharmacogenomics, 16S rRNA microbiome sequencing, and circulating tumor DNA to name a few.

Catalina Lopez-Correa, MD, PhD

Canada

Chief Scientific Officer, Genome Canada

Presently, Dr. Lopez-Correa is the Chief Scientific Officer (CSO) at Genome Canada. As CSO, she is taking genomics to the next level, by advancing mission driven initiatives using genomics to solve the most pressing global challenges. Previously, she was Executive Director of the Canadian COVID19 Genomics Network (CanCOGen), a $40M initiative to advance the use of genomics to help us understand and control the COVID19 pandemic. She also held the position of COO at Ruta N Medellin, driving innovation ecosystems to advance social and economic development in Colombia and Latin America. Prior to that, Dr. Lopez-Correa was the CSO at Genome BC and at Genome Quebec where she was instrumental in developing competitive teams for national and provincial research and innovation initiatives raising the profile of Canadian genomics on the global stage. Dr. Lopez-Correa has also held leadership positions at deCODE Genetics, and Eli Lilly and has played advisory roles at the European Commission, Innovative Medicine Initiatives and other private and public sector entities working in the application and implementation of genomics technologies. As part of her commitment to international development, Dr. Lopez-Correa has led several initiatives aimed at demonstrating the impact of genomics and innovation in developing countries. She is also fully committed to advance equity, diversity and inclusion, with a particular interest in gender equity in science.
In 2017 she received the Canadian Senate 150th Anniversary Medal. In 2013 she was recognized by National Order of Merit Award in the Rank of Officer from the Republic of Colombia.

Dr. Lopez-Correa holds an MD from the Universidad Pontificia Bolivariana in Colombia, an MSc in Human Genetics from Paris V University in France, a PhD in Medical Sciences from the KULeuven in Belgium, and a mini MBA from McGill University in Canada.

Megan Lynch

USA

PhD Student, University of Maryland, Baltimore

I am a PhD candidate in the Genetics and Genomic Medicine program at the University of Maryland School of Medicine. My research includes studying the genetic architecture of a founder population including statistical analysis of disease-susceptibility genes in an Amish community, evaluating the association of reproductive outcomes with genetic relatedness of couple pairs, mapping autozygosity and complex traits, and mapping epigenetic modification in familial hyperlipidemia carriers and postprandial hyperlipidemia.

Vinicius Maracajá-Coutinho

Chile

Assistant Professor, Universidad de Chile

Dr. Maracaja-Coutinho is a Group Leader at the University of Chile, Associate Researcher of the Advanced Center for Chronic Diseases - ACCDiS, founder of Beagle Bioinformatics, Director of the Graduate Diploma in Bioinformatics and Computational Biology from the University of Chile, and President of the Chilean Bioinformatics Society. His research is focused in data-driven genomics to achieve the functional aspects, evolution and disease association of small and long non-coding RNAs, through the integration and application of: (i) computational systems biology; (ii) RNA bioinformatics; (iii) comparative genomics; (iv) epitranscriptomics of diseases; (v) single-cell transcriptomics; and (vi) development of computational tools/databases for genomic data analysis.

Sonia Margarit, MS, CGC

Chile

Genetic Counselor, Universidad del Desarrollo

Sonia Margarit is a certified genetic counselor. She graduated in 1997 from the Human Genetics Program at Sarah Lawrence College in Bronxville, New York, and subsequently worked for seven years in the prenatal and pediatric areas at Elmhurst Hospital / Mount Sinai Hospital in New York.

In 2004, she moved to Santiago, Chile, and became Chile's first genetic counselor at the Clínica Alemana Universidad del Desarrollo at the Center for Genetics and Genomics and became an associate professor at the Faculty of Medicine.

In 2014 she obtained her master's degree in Health Psychology at the Catholic University of Chile. Besides her academic and clinical work, she is a passionate promoter of the importance of formal training in genetic counseling. For the last 5 years, recognizing the lack of training courses among healthcare providers, she is involved in the development of diplomas and certificate courses in hereditary cancer and genetic counseling.

Emmanuel Jairaj Moses, BSc, MSc, PhD

Malaysia

Senior Lecturer, Universiti Sains Malaysia

Alison Motsinger-Reif, PhD

USA

Branch Chief, National Institute of Environmental Health Sciences

Alison Motsinger-Reif, Ph.D., is Chief of and a principal investigator in the Biostatistics and Computational Biology Branch. Dr. Motsinger-Reif earned both an MS in Applied Statistics and a PhD in Human Genetics from Vanderbilt University. She was a faculty member at North Carolina State University from 20017-2018. Overall, her group focuses on the development and application of modern statistical approaches for understanding the etiology of common, complex diseases. As the field of human genetics increasingly accepts a complex model of phenotypic development involving many genetic and environment factors, it is increasingly important to develop analytical strategies that incorporates this complexity.

Maximilian Muenke, MD, FACMG

USA

CEO, American College of Medical Genetics and Genomics

Recognized as a highly acclaimed physician-scientist and dedicated clinical and research mentor, Dr. Muenke brings more than three decades of experience to the ACMG, including 10 years on the faculty of the University of Pennsylvania School of Medicine. Since 2000, Dr. Muenke has served as Senior Investigator, Head of the Human Development Section, and Chief of the Medical Genetics Branch at the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH). The focus of his laboratory's research was on the delineation and identification of the underlying causes of craniofacial and related anomalies in humans including holoprosencephaly as well as Muenke syndrome, a relatively common craniosynostosis syndrome. Dr. Muenke is passionate about training the next generation of leaders in the field of genetics and genomic medicine. He has directed medical genetics training since 1994, first at the University of Pennsylvania and then at the NIH, where he was the Director of the NIH Medical Genetics and Genomic Medicine Residency and Fellowship Training Programs.

Dr. Muenke earned his MD degree at the Free University of Berlin School of Medicine, Germany in 1979. After his residency in pediatrics at the Christian-Albrechts University in Kiel, Germany and a postdoctoral fellowship in human genetics at Yale University, he completed a clinical genetics fellowship at the Children's Hospital of Philadelphia. He is board certified in clinical genetics, clinical cytogenetics and clinical molecular genetics by the American Board of Medical Genetics and Genomics.

Among his many accomplishments and honors, Dr. Muenke has received merit awards from the NHGRI and the NIH Office of the Director—both recognizing his work in the training of future geneticists—three NHGRI GREAT (Genome Recognition Employee Accomplishments and Talents) Awards, the Lifetime Achievement Award in Medical Genetics from the African Society of Human Genetics and the NIH Director's Award for establishing The International Summit in Human Genetics and Genomics. In 2019, Dr. Muenke was awarded the Samuel Pruzansky Memorial Lecture Award from the March of Dimes Birth Defects Foundation.

Education:
1972 Abitur Goethe-Gymnasium, Ibbenbüren, Germany
1979 M.D. Free University of Berlin School of Medicine
M.D. Thesis Title: Silver staining of nucleolus organizer regions on human prematurely condensed chromosomes from cells with different ribosomal RNA gene activity. (Magna cum laude in Human Genetics).

Postgraduate Training and Fellowship Appointments (2005-2012 only):
2005 Medical Acupuncture (300 hours), New York Medical College, Department of Community and Preventive Medicine (Program Director: Ravinder Mamtani, M.D.)
2006 Introduction to Clinical Hypnosis (20 hours), Greater Philadelphia Society of Clinical Hypnosis

Amy Nisselle, PhD

Australia

Genomics Workforce Lead, Specialist Project Officer, Melbourne Genomics, Murdoch Children's Research Institute

Dr Amy Nisselle leads the Genomics Workforce stream of Melbourne Genomics, investigating the genomics practice and educational needs of the clinical, diagnostic and data science workforces. She is also a Specialist Project Officer, Genomics in Society, Murdoch Children's Research Institute, and an Honorary Senior Fellow of The University of Melbourne. Amy has a doctorate in multimedia genetics education, and 20 years' experience in medical, science and education research in Australia, the USA and UK. Amy's current research focuses on education and training of health professionals in the genomics era, based in contemporary theories of adult education, behaviour change and evaluation. She uses research insights to inform multifaceted genomics education and training programs and determine effective strategies to develop the workforce. Through her work with Australian Genomics she creates tools to support effective genomics education and build an evidence base of best practice. Amy convenes the Genomic Education Network of Australasia, is Co-Chair of the Human Genetics Society of Australasia's Education, Ethics and Social Issues Committee, and a member of the Global Genomics Medicine Collaborative (G2MC) Education Working Group and the National Institutes of Health's Inter-Society Coordinating Committee on Practitioner Education in Genomics.

Lori Orlando, MD, MHS, MMCI

USA

Director Program in Precision Medicine, Duke

Dr. Lori A. Orlando, MD, MHS, MMCI, is a Professor of Medicine, health services researcher, and Director of the Precision Medicine Program in the Center for Applied Genomics and Precision Medicine at Duke University. She received her MD from Tulane University in 1998, MHS from Duke in 2004, and MMCI from Duke in 2019. She completed her fellowship, specializing in decision modeling and technology assessments, in 2004. Her research expertise is in decision modeling and implementation science as relates to identifying and managing individuals in clinical settings at increased risk for medical conditions. Her current research program focuses upon using technology to overcome barriers

to family health history based risk assessment and using high quality family health histories to guide clinical care.

George Patrinos

Greece

Professor and Head of Laboratory, University of Patras, School of Health Sciences, Department of Pharmacy

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, since 2010, he is Full Member and Greece's National representative in the CHMP Pharmacogenomics Working Party of the European Medicines Agency (EMA, Amsterdam, the Netherlands) and since 2018 Co-Chair of the Global Genomic Medicine Collaborative (G2MC).

George is currently Head of the Laboratory of Pharmacogenomics and Individualized Medicine, the first officially established academic laboratory on pharmacogenomics in Greece. His group consists of more than 35 staff members from graduate students to post-doctoral scientists, covering disciplines from wet and dry lab and public health genomics projects, all focusing on pharmacogenomics and personalized medicine. In particular, his research interests involve discovery work and clinical implementation of pharmacogenomics, focusing in particular in psychiatry but also cardiology and oncology, genomics of rare disorders and transcriptional regulation of human fetal globin genes. Moreover, George's group is internationally recognized for its involvement in developing National/Ethnic Genetic databases to document the genetic heterogeneity in different populations worldwide and of genome informatics tools to translate genomic information into a clinically meaningful format. Also, George's group has a keen interest in public health genomics to critically assess the impact of genomics to society and public health.

George has more than 270 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. Also, he 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. Furthermore, he serves as Editor-In-Chief of the prestigious Pharmacogenomics Journal (TPJ), published by Nature Publishing Group, Associate Editor and member of the editorial board of several scientific journals, such as Human Mutation, Human Genetics, Human Genomics, Pharmacogenomics, etc and has been a member of several international boards and advisory and evaluation committees.

Apart from that, George is the main co-organizer of the Golden Helix Conferences, an international meeting series on Pharmacogenomics and Genomic Medicine.

Eduardo Pérez-Palma, PhD

Chile
University del Desarrollo

Eduardo did his PhD in Santiago de Chile where he studied the genetics of the Chilean population. He continued with a Postdoctoral fellowship at the Cologne Center for Genomics in Cologne, Germany and then worked as a research associate at the Genomic Medicine Institute of Cleveland Clinic, Cleveland USA. To date he has published more than 30 manuscripts focusing on epilepsy genetics and bioinformatics. Overall, his work aims to bridge novel genetic knowledge with clinical practice. Since January, Eduardo joined the Centro de Genética y Genómica of Universidad del Desarrollo as a Principal Investigator.

Alexandra Pickard

United Kingdom

Deputy Director, Genomics, NHS England and NHS Improvement

Alexandra Pickard is a Deputy Director in the Genomics Unit in NHS England and NHS Improvement. The Genomics Unit is responsible for supporting the delivery of the NHS Genomic Medicine Service for the NHS in England. The NHS Genomic Medicine Service consists of a network of NHS Genomic Laboratory Hubs, an integrated clinical genomics service and NHS Genomic Medicine Service Alliances supporting the embedding of genomic testing in end to end clinical pathways.

Alexandra is responsible for leading the policy and strategy development for the NHS GMS, as well as overseeing the commissioning and finance arrangements for the service.

Prior to joining NHS England and NHS Improvement Alexandra worked in healthcare policy consultancy.

Gabriel Rada, MD

Chile

Director, Epistemonikos Foundation

Gabriel Rada is the co-founder, president and CEO of Epistemonikos foundation (asociación sin fines de lucro “Epistemonikos”) a not-for-profit organization which mission is to bring independent, high-quality information closer to everyone making health decisions. He leads projects in Evidence-Based Health care and is currently working in technologies that combine machine and human collaborative efforts to generate knowledge, multilingual platforms for decision-makers, and automation of systematic reviews.

Boris Rebolledo-Jaramillo, PhD

Chile

Assistant Professor, Universidad del Desarrollo

Boris Rebolledo-Jaramillo is an Assistant Professor at the Institute for Science and Innovation in Medicine at Universidad del Desarrollo, Chile. He received his B.Sc. in Bioengineering (2009) and M.Sc. in Bioinformatics (2012) from University of Concepcion, Chile, and Ph.D. (2016) in Bioinformatics and Genomics from Penn State University.

Dr. Rebolledo-Jaramillo's work has focused on the development and implementation of bioinformatics methods for the analyses of sequencing data, with an emphasis on mitochondrial genomics. Currently, his work is focused on clinical genomics. Particularly, the clinical consequences of variants affecting the mitochondrial-nuclear genetic coordination, and the implementation of exome sequencing analyses for the diagnostic of rare disorders.

Heidi Rehm, PhD

USA

Chief Genomics Officer and Professor of Pathology, Massachusetts General Hospital and Broad Institute of MIT and Harvard

Heidi Rehm is the Chief Genomics Officer in the Department of Medicine and at the Center for Genomic Medicine at Massachusetts General Hospital working to integrate genomics into medical practice. She is a board-certified laboratory geneticist, Medical Director of the Broad Institute Clinical Research Sequencing Platform and Professor of Pathology at Harvard Medical School, working to guide genomic testing for clinical and clinical research use. She is a principal investigator of ClinGen, providing free and publicly accessible resources to support the interpretation of genes and variants. Rehm also co-leads the Broad Center for Mendelian Genomics focused on discovering novel rare disease genes and co-leads the Matchmaker Exchange to also aid in gene discovery. She is a strong advocate and pioneer of open science and data sharing, working to extend these approaches through her role as vice chair of the Global Alliance for Genomics and Health. Rehm is also a principal investigator of the Broad-LMM-Color All of Us Genome Center supporting the sequencing and return of results to a cohort of one million individuals in the US and co-leading gnomAD, the Genome Aggregation Database.

Gad Rennert, MD, PhD

Israel

The Chil and Berta Weissman Professor of Precision Medicine, Technion, and Chairman, Dept of Communi, Technion-Israel Institute of Technology and Clalit Health Services

I am the Chil and Berta Weissman Professor of Precision Medicine and Chairman, Dept. Community Medicine and Epidemiology, Carmel Medical Center and B. Rappaport Faculty of Medicine, Technion-Israel Institute of Technology and Director, Clalit National Cancer Control Center and National Personalized Medicine Program. I earned my M.D. degree from Ben-Gurion University School of Medicine, Beer Sheva, Israel and my Ph.D. Degree from the School of Public Health, University of North Carolina Chapel Hill. I am a molecular epidemiologist with a particular interest in cancer predisposition, and gene-environment/diet interactions. I have more than 350 publications on genetics and environmental causation of colorectal, breast, lung, gynecological, pancreato-hepato-biliary and other cancers, mostly based on large-scale case-control studies with some 50,000 participants which I am running. In addition, I am coordinating population cancer screening activities on a national level in Israel (from policy to implementation-adherence, quality and outcomes), and studying unique founder mutations of cancer risk through the Familial Cancer Consultation Service which cares for thousands of mutation carriers.

Gabriela Repetto, MD

Chile

Director, Rare Diseases Program, Centro de Genetica y Genomica Facultad de Medicina, Clinica Alemana Universidad del Desarrollo

Professor of Genetics, Facultad de Medicina, Clinica Alemana Universidad del Desarrollo in Santiago, Chile. Former President (2018-2020), Sociedad de Genetica de Chile. Member of the Board of Directors, Global Genomic Medicine Collaborative. I work as a clinician, educator and researcher in Human and Medical Genetics and Genomics, with a special focus on rare disorders.

Emma Rey-Jurado, PhD

Chile

Professor Researcher, University del Desarrollo

- Biologist (Universidad Autónoma de Barcelona, Spain) 2007
- PhD on Biomedicine (Universidad de Barcelona, Spain) 2013
- National Institute for Medical Research, London, UK, 2013
- Post-doctoral Scientist, Public Health Research Insititute (PHRI), Rutgers, 2015
- Post-doctoral Scientist, Pontificia Universidad Católica de Chile, Chile, 2015-2018
- Professor Researcher at Universidad del Desarrollo, Facultad de Medicina, Instituto de Ciencias e Innovación en Medicina, 2020 - Nowadays
- Research interests: Primary Immunodeficiencies, Susceptibility to infections, autoimmunity and autoinflammation.

Charles Rotimi, PhD

USA

NIH Distinguished Investigator, Metabolic, Cardiovascular and Inflammatory Disease Genomics Branch, NHGRI

Charles Rotimi, a genetic epidemiologist, is an NIH Distinguished Investigator and the Director of the Trans-NIH Center for Research on Genomics and Global Health. He is a leader in exploring the implications of the increased genetic diversity in African ancestry populations for disease gene mapping. Rotimi is especially proud of his efforts at globalizing genomics. His engagement of African communities for the International HapMap and 1000 Genomes projects has had a transformative impact. Rotimi was the founding president of the now thriving African Society of Human Genetics, and spearheaded formation of the H3Africa Initiative with over 176 million US dollars funding from NIH and Wellcome Trust. Rotimi was recognized as an “African Innovator” by Quart Africa and elected to the USA National Academy of Medicine and African Academy of Sciences. He is 2020 president-elect for the American Society of Human Genetics. He is an internationally recognized scholar with research accomplishments profiled in leading international scientific journals and news media including New-England-Journal of Medicine, Science, Nature, Lancet, Newsweek, NY Times and BBC.

Brock Schroeder, PhD

USA

Senior Director, Global Market Access Strategy, Illumina

Brock Schroeder is Senior Director of Global Market Access Strategy & HEOR at Illumina, where he leads efforts to generate evidence of clinical and economic utility of current and emerging clinical applications of NGS to support HTA, payer coverage, and patient access to clinical genomic tools. Prior to joining Illumina in 2017, Brock led Medical Affairs at Biotheranostics.

Brock's industry experience includes multiple therapeutic areas, including oncology, rare diseases, and reproductive health. Brock has authored >25 peer-reviewed manuscripts and >50 scientific congress abstracts. He received his BA in Biology from Washington University, and his PhD in Neuroscience from University of Wisconsin-Madison.

Nirmala Sirisena, PhD

Sri Lanka

Senior Lecturer and Clinical Geneticist, Human Genetics Unit, Faculty of Medicine, University of Colombo

Dr. Nirmala D. Sirisena's special interests are Clinical Genetics, Cancer Genetics and Genomics, Genomic Medicine and Genomic Education. She provides clinical care and genetic counselling services at the Human Genetics Unit. She is also involved in teaching clinical genetics and genomics to both undergraduate and postgraduate students in the Faculty and co-ordinating research work related to both clinical genetics and cancer genomics.

She spearheads the cancer genetics program in the Unit and has been involved in integrating next generation sequencing based cancer gene panel testing to the repertoire of services offered through the clinic. Her current research work involves characterizing the molecular genetic determinants of breast and colorectal cancer in the Sri Lankan population. She is also actively involved in promoting genomic medicine training programs for healthcare workers in the country. She is a Steering Committee member of the G2MC initiative and a Co-chair of the Young Investigator Sub-committee.

Tai E Shyong

Singapore

Professor, National University of Singapore

Professor Tai is an endocrinologist and clinician scientist who has been involved in numerous large-scale genetic association studies related to diabetes and metabolic disease. He is also the Chief Medical Officer of PRECISE, the organisation that oversees Phase II of the national precision medicine programme in Singapore.

Meow-Keong Thong, MBBS, MPaeds, MD, FHGSA (Clinical Genetics), FAMM, FASc

Malaysia

Head, Genetics & Metabolism Unit, Department of Pediatrics & Head, Genetic Medicine Unit, University of Malaya Medical Centre

Dr THONG Meow-Keong is a Professor of Paediatrics and Consultant Clinical Geneticist at the University of Malaya Medical Centre. He was a Fulbright Scholar and a board-certified clinical geneticist and established the first Genetics Clinic in Malaysia in 1995. He is the current President of the College of Paediatrics, Academy of Medicine of Malaysia; Vice-President of the Medical Genetics Society of Malaysia, Trustee of the Rare Disease Alliance Foundation Malaysia, appointed member of the Malaysia Medical Council (Education) and Advisor to the Malaysian Rare Disorders Society. He was the Head, Department of Paediatrics, University of Malaya and past President, Asia-Pacific Society of Human Genetics.

His clinical practice and research are focused on rare diseases, genomic medicine, inborn errors of metabolism and genetic counselling. He has published extensively in the field of paediatrics and genetic medicine in low-resource settings. He has authored/co-authored over 100 WoS/ISI journal publications, 3 books, 18 book chapters including the Oxford Monograph in Medical Genetics and an

IDEAS White Paper entitled “Rare Diseases in Malaysia”. He was consulted by the World Health Organization and the Ministry of Health Malaysia on various technical issues and clinical practice guidelines. He was active in developing undergraduate and postgraduate paediatric training curriculum programs and promoted advocacy issues affecting children and individuals with rare diseases. He has won major research awards and research grants and has collaborations with major universities and non-governmental organizations globally. He was elected a Fellow of the Academy of Sciences Malaysia, Academy of Medicine of Malaysia and Academy of Medicine, Singapore.

Jason Vassy, MD, MPH, MS

USA

Associate Professor, Harvard Medical School at VA Boston Healthcare System

Dr. Vassy is an Associate Professor of Medicine at Harvard Medical School, a clinician-investigator at the VA Boston Healthcare System (VABHS) and Brigham and Women’s Hospital (BWH), and a practicing primary care physician. He directs the Genomes2Veterans research program, whose research examines the clinical utility of genetic and genomic testing in various primary care clinical contexts. Current projects include clinical trials of pharmacogenetic testing, polygenic risk scores, and return of unanticipated genetic results.

Tania Vasquez-Loarte, MD, MPH

USA

Health Coach, Wyckoff Heights Medical Center

I am a public health geneticist who is passionately committed to the improvement of the gaps in the healthcare of children with genetic conditions in emerging countries. My passion for public health genetics started while participating in medical missions, where I witnessed the need for medical genetics care in underserved communities. My experience includes the implementation and evaluation of newborn screening, assessment of healthcare gaps in rare disorders, the evaluation of patients' values and bioethical analysis in the implementation of genetic technologies, the elaboration of clinical guidelines, measuring health outcomes from patient registries and policy making to regulate human research. I am currently based in NY but am originally from Peru. Like many Peruvians, I love cooking, spending time with my family and I am constantly learning traditional Peruvian dances. I also love photography.