GLOBAL COMMISSION | to End the Diagnostic Odyssey for Children with a Rare Disease

Co-Chair & Members List

COMMISSION CO-CHAIRS

Simon Kos, M.D.
Chief Medical Officer & Sr. Director Microsoft Worldwide Health

Yann Le Cam
Chief Executive Officer, EURORDIS–Rare Diseases Europe

Flemming Ornskov, M.D., M.P.H.
Chief Executive Officer
Shire

COMMISSION MEMBERS

Moeen Al-Sayed
Chairman of Medical Genetics
King Faisal Specialist Hospital

Kym Boycott
Clinical Geneticist, Children's Hospital of Eastern Ontario
Senior Scientist, CHEO Research Institute
Professor, Department of Pediatrics, University of Ottawa

Roberto Giugliani
Medical Genetics Service, Hospital de Clinicas de Porto Alegre
Professor of Medical Genetics, Federal University of Rio Grande do Sul, Brazil

Kevin Huang
President
Chinese Organization of Rare Disorders

Derralynn Hughes
Clinical Director Haematology Oncology and Palliative care, Senior Lecturer and Investigator Lysosomal Storage Disorders Unit, Royal Free & University College Medical School

Daniel MacArthur
Institute Member, Co-Director of the Medical and Population Genetics Program
Broad Institute

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Maryam Mohd. Fatima Matar
Founder and Chairperson
UAE Genetic Diseases Association

Dau-Ming Niu
Director, Center for Medical Genetics
Taipei Veterans General Hospital

Mike Porath
Founder and Chief Executive Officer
The Mighty

Arndt Rolfs
Chief Executive Officer
Centogene

Richard Scott
Clinical Lead
Rare Disease, 100,000 Genomes Project at Genomics England
Consultant and Honorary Senior Lecturer in Clinical Genetics, Great Ormond Street Hospital for Children and the UCL Institute of Child Health

Marshall Summar
Director, Rare Disease Institute; Division Chief, Genetics and Metabolism
Children's National

Durhane Wong-Rieger
President and Chief Executive Officer
Canadian Organization for Rare Disorders
Council Member
Rare Diseases International

CO-CHAIR & MEMBER INFORMATION

Simon Kos, M.D.

Dr. Kos is Chief Medical Officer and Senior Director, Worldwide Health, Microsoft. In this key executive role, Dr. Kos is responsible for providing clinical guidance, thought leadership, vision and strategy for Microsoft technologies and solutions in the Health and Healthcare industries. He works with industry partners and healthcare organizations around the world to improve and transform health outcomes by leveraging technology and innovation.

Dr. Kos joined Microsoft in 2010, bringing his expertise to the Australian health team after 17 years in the health and health IT industries. As the doctor on Microsoft's health team, he was responsible for health strategy and industry engagement. He raised awareness of the Microsoft brand, technologies, and partner community, and he works to find clinical relevance for Microsoft products.

Prior to coming to Microsoft, Dr. Kos worked with global health IT companies Cerner and InterSystems as they implemented some of the largest e-Health initiatives in Australia. His responsibilities included product design and localization, clinical engagement, industry thought leadership, and team management. Before his career in health IT, Dr. Kos practiced at NSWHealth for several years with a focus on critical care.

Dr. Kos has an MBBS and a BSc (Med) from the University of New South Wales, as well as an MBA with a major in change management from the Australian Graduate School of Management. He is passionate about improving healthcare through technology.

Yann Le Cam

Mr. Le Cam is a patient advocate who has dedicated 25 years of professional and personal commitment to health and medical research non-governmental organizations in France, Europe and the United States in the fields of cancer, HIV/AIDS and rare diseases.

He has three daughters, the eldest of whom is living with cystic fibrosis. Mr. Le Cam is one of the founders of EURORDIS in 1997 and the organization's Chief Executive Officer since 2001.

Yann Le Cam has participated in the revision and adoption of European regulations that impact the lives of rare disease patients, including the EU

Regulation on orphan medicinal products.

He was one of the first patient representatives appointed to the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency (EMA), where he served for 9 years and was its vice-chair for 6 years. He served on the Management Board and Executive Committee of the French HTA agency for 5 years, on the DIA Advisory Committee Europe for 3 years.

He was the Vice Chairman of the EU Committee of Experts on Rare Diseases (EUCERD) from 2011 to July 2013, and he is nominated on the current Commission Expert Group on Rare Diseases.

Mr. Le Cam is also a member of and immediate past Chair of the Therapies Scientific Committee of IRDIRC (the International Rare Diseases Research Consortium).

In June 2016, Mr. Le Cam was elected to the Management Board of the European Medicines Agency.

Mr. Le Cam is a founder and a member of the Council of Rare Diseases International.

Flemming Ornskov, M.D., M.P.H.

Dr. Ornskov brings to his role his operational and medical knowledge and his extensive international, strategic and operational experience in the pharmaceutical sector. He formerly held the position of Non-Executive Chairman of Evotec AG and was Non-Executive Director of PCI Biotech Holding ASA. From 2010 to 2012 he was Chief Marketing Officer and Global Head, Strategic Marketing for General and Specialty Medicine at Bayer. From 2008 to 2010 Dr. Ornskov served as Global President, Pharmaceuticals and OTC at Bausch & Lomb, Inc. He also served as Chairman, and later as President and Chief Executive Officer, of Life-Cycle Pharma A/S from 2006 to 2008, and as President and Chief Executive Officer of Ikaria, Inc. from 2005 to 2006. Earlier in his pharmaceutical career Dr. Ornskov held roles of increasing responsibility at Merck & Co., Inc. and Novartis AG, following a distinguished period spent in hospitals and academic medicine. Dr. Ornskov received his M.D. from the University of Copenhagen, MBA from INSEAD and Master of Public Health from Harvard University.

Dr. Ornskov is a member of the Swiss-American Chamber of Commerce (Non-Executive Director) and Waters Corporation (Non-Executive Director).

Moeen Al-Sayed, M.D.

Prof. Al-Sayed is Chairman of the Department of Medical Genetics at King Faisal Specialist Hospital, and Professor of Genetics at Al-Faisal University in Riyadh, Kingdom of Saudi Arabia. Professor Al-Sayed is also the Director of the Genetic Counselling Service at King Faisal Specialist Hospital and MSc Genetic Counselling Programme at Al-Faisal University. He has established a Postgraduate Genetic Counselling Programme at King Faisal Specialist Hospital to train Saudi and Arab nationals in this field and to promote this specialty across the Middle East.

He obtained his Bachelor's degree in Medicine and Surgery (MBBS) from the Faculty of Medicine, King Saud University, Riyadh. He completed his paediatric residency at Yale–New Haven Children's Hospital, and gained his fellowship in clinical genetics and clinical biochemical genetics at Baylor College of Medicine, Houston, TX, USA. Prof. Al-Sayed has an MBA from the University of Tennessee, Knoxville, TN, USA.

His interests include identification of founder mutations related to metabolic and genetic disorders in the Saudi population, screening and treatment of lysosomal storage diseases (LSDs) and the management of organic acidurias. Prof. Al-Sayed is a founding member of the Middle Eastern Metabolic Group (MEMG), the Saudi Pediatric Association – Medical Genetics Subcommittee, the Saudi Charitable Society for Genetic Disorders, the Saudi Society of Medical Genetics and the Middle Eastern Lysosomal Storage Diseases Expert Council (MELSDEC).

Kym Boycott, M.D., Ph.D., FRCPC, FCCMG

Kym Boycott is a Clinical Geneticist at the Children's Hospital of Eastern Ontario (CHEO), Senior Scientist at the CHEO Research Institute, and Professor of Pediatrics at the University of Ottawa. Dr. Boycott's research program in rare diseases bridges clinical medicine to basic research and is focused on understanding the molecular pathogenesis of these disorders to improve patient care and family well-being. She is the Principal Investigator of Care4Rare Canada, a pan-Canadian platform integrating genomic and other –omic technologies to improve our understanding of rare disease, with a particular focus on solving the unsolved and most difficult rare diseases. She is co-Principal Investigator of the Rare Diseases: Models & Mechanisms Network, established to catalyze connections between clinical investigators discovering new genes and basic scientists who can analyze equivalent genes and pathways in model organisms. Dr. Boycott moves the international rare disease agenda forward through her role as the

Chair of the Diagnostics Committee of the International Rare Diseases Research Consortium and member of the Steering Committee of the Global Alliance for Genomics and Health.

Roberto Giugliani, M.D., Ph.D., M.Sc.

Roberto Giugliani is a Medical Doctor, with specialization in Medical Genetics. He obtained his MD degree at UFRGS, in Porto Alegre, Brazil, the PhD degree at USP, in Ribeirão Preto, Brazil, and had post-doctoral trainings in London, Genoa, Paris, Zurich, Oakland and Sydney. He is Professor of the Department of Genetics at UFRGS, Coordinator of the National Institute of Population Medical Genetics (INAGEMP) and Director of the WHO Collaborating Center for the Development of Genetic Services in Latin America, in Porto Alegre, Brazil. He founded the Medical Genetics Service of Hospital de Clínicas de Porto Alegre, where he is still an active member. He acted as President of the Brazilian Society of Clinical Genetics (SBGM), of the Latin American Network of Human Genetics (RELAGH) and of the Latin American Society of Inborn Errors of Metabolism and Neonatal Screening (SLEIMPN). He currently is Researcher Level I A (top) of the Brazilian Research Council (CNPq), is member of 11 scientific societies (including the Brazilian Academy of Sciences) and is the Editor-in-Chief of the Journal of Inborn Errors of Metabolism and Screening. His main research interest is concentrated in the field of inborn errors of metabolism, particularly the lysosomal disorders, and has over 450 papers published in peer-reviewed scientific journals.

Kevin Huang

Kevin Huang graduated from Zhejiang University City College. He is the founder and president of the Chinese Organization for Rare Disorders. He is also a patient with a rare disease. Kevin was the one who brought “International Rare Disease Day” to China. Through his work, rare diseases are now widely known in China. Kevin has also been a champion promoting communications and facilitating collaborations among various rare disease stake-holders. He founded the China Rare Disease Patient Organization Network and started the China Rare Disease Summit – the most influential rare disease conference in China. He is a pioneer and practitioner and has become an acknowledged iconic figure in rare disease field in China.

Derralynn Hughes, Ph.D.

Dr. Hughes trained in medicine at Oxford University during which she intercalated a doctorate in macrophage physiology. After training in Haematology in London she was appointed to lecturer in haematology with a special interest in lysosomal storage disorders at the Royal Free Hospital through UCL and subsequently to Senior Lecturer and consultant in 2008. She has initiated and led a comprehensive clinical trials program for LSDs, in particular Fabry disease. Dr. Hughes has an international reputation as a principal investigator having been awarded NIHR principal leading principal investigator awards in 2015 and 2016. She has developed a laboratory research program successfully supervising Ph.D., M.D. and M.Sc. students to thesis submission and award with the laboratory producing contributions in the area of bone pathophysiology in Gaucher disease and has recently described for the first time the increased incidence of malignancy in that Fabry disease. She is an author of over 160 papers in the area of macrophage biology and lysosomal Storage Disorders. Within the hospital she is Clinical Director of Haematology, Oncology and Palliative care and lead cancer physician. She has led a range of initiatives to improve cancer patients' outcomes and experience and has designed, lead and initiated a new academic program for multidisciplinary training of health care professionals in cancer.

Daniel MacArthur, Ph.D.

Dr. MacArthur is an institute member at the Broad Institute of MIT and Harvard, and co-director of the Broad's Program in Medical and Population Genetics. In addition to his roles at the Broad, Dr. MacArthur is a group leader in the Analytic and Translational Genetics Unit at Massachusetts General Hospital and an assistant professor at Harvard Medical School. Dr. MacArthur's lab seeks to accurately identify DNA variants affecting gene function and human disease risk from large-scale datasets containing genetic information from more than 90,000 human genomes. His research brings these approaches together to discover disease-causing mutations in patients with rare severe diseases, with a particular focus on neuromuscular diseases such as muscular dystrophy. He has already used these techniques to diagnose several patients, and is working with collaborators to design genetic tests and treatments for the patients and their families. Dr. MacArthur completed his Ph.D. at the Institute for Neuromuscular Research in Sydney, Australia, where he studied a loss-of-function variant in the human ACTN3 gene associated with variation in muscle strength and athletic performance. He later served as a postdoctoral

fellow at the Wellcome Trust Sanger Institute in Hinxton, UK, where he led the annotation of gene-disrupting ("loss-of-function") variants as part of the 1000 Genomes Project Consortium.

Maryam Mohd. Fatima Matar, M.D.

Dr. Matar is among the "top 100 most influential Arabs in the world," recognized by Arabian Business 2017. She is recognized as the most powerful scientist in the UAE since 2014 and among the top 20 Arab scientists with the biggest contribution to humanity by British Scientific Community 2016. Dr. Matar is the first Emirati woman appointed as Director General, of the Community Development Authority (CDA) and the first Emirati woman in the position of Undersecretary to the Ministry of Health in 2006. She is a women in STEM (Science Technology, Engineering and Mathematics) Ambassador and advisor on Women Leadership in workplace. Dr. Matar is a leading influencer advising private sector organizations, healthcare and educational institutions, on their strategy regarding the sustainable development. She is also a globally acclaimed speaker on the topics of emerging technologies, biotechnology, epigenetics, impact of genetic screening and leadership.

Dau-Ming Niu, M.D., Ph.D.

Dr. Dau-Ming Niu is a professor in Institute of Clinical Medicine at National Yang-Ming University and director of Genetic and Endocrinology Division, Research and Treatment Center of Rare Disease at Taipei Veterans General Hospital.

Dr. Niu received his M.D. from Kaohsiung Medical University, was a clinical and research fellow at National Taiwan University Hospital. He had advanced studies in Professor Yuan-Tsong Chen's laboratory in medical genetics at Duke University. After coming back to Taiwan, Dr. Niu received a Ph.D. degree from National Yang-Ming University of Clinical medicine.

Dr. Niu's research interests include medical genetics and endocrinology. His research in 6-pyruvoyl-tetrahydropterin synthase deficiency has the most outstanding treatment outcome, ranked No. 1 in the world so far. In addition, his team found that Taiwanese aboriginals have the highest prevalence of homocysteinuria and inherited Retinitis Pigmentosa across the globe. In recent years, Dr. Niu has focused his studies intensively on lysosomal storage diseases and his team is the first to discover that Taiwan has the highest prevalence of Fabry disease

in the world. During this period of time, a research and treatment center of rare disease was also established, which is the first center integrating research and medical expertise of different areas in the rare disease field in Asia.

Mike Porath

Mike Porath is the founder and CEO of The Mighty, the leading digital health community that reaches more than 20 million people a month.

Mike's entry into the rare disease community came through his daughter, who has Dup15q syndrome, a neurogenetic disease associated with autism spectrum disorder, intellectual disability, and epilepsy.

Soon after her diagnosis, Mike joined the board of Dup15q Alliance and now also serves as fundraising chair. The community he became a part of there inspired him to create The Mighty to empower and connect people facing rare diseases and other health conditions.

Mike spent most of his career as a journalist, where he won multiple awards and held a variety of reporting, editing, producing and executive roles at media companies including ABC News, NBC News, The New York Times and AOL.

He has become a leading voice for patient-centered healthcare and speaks at events around the world about leveraging people and technology to improve the lives of patients.
Mike also serves as an advisor to the UCLA Neurogenetics Clinic and as a mentor to Cedars-Sinai Accelerator powered by Techstars, where he helps technology innovators turn their ideas into breakthroughs.

He is proud and honored to serve on this commission.

Arndt Rolfs, M.D.

Arndt Rolfs, M.D., Professor for Neurology and Psychiatry, is appointed CEO and founder of Centogene AG, and Director of the Albrecht-Kossel-Institute for Neuroregeneration at the University of Rostock, Germany. He received his

approbation for human medicine in 1985 from the University of Mainz, Germany and the University of Vienna, Austria. Arndt is a principle investigator of several international multicentre studies in the area of rare diseases and actively engaged in biomarker research for several metabolic diseases, including the Sifap project (www.sifap.eu), the world's largest study in young stroke patients related to Fabry disease, several biomarker studies (e.g. BioHAE, BioGaucher, BioHunter, BioMorquio) and epidemiological studies in neurogenetic aetiologies. Arndt has an extensive track record in medical and scientific publications with more than authored/contributed to 250 peer-reviewed publications and acts as a consultant for international biotech and industrial companies, and is also a consultant for the EMA and FDA.

Richard Scott, Ph.D.

Dr. Scott is the Clinical Lead for Rare Disease for the 100,000 Genomes Project at Genomics England and a Consultant and Honorary Senior Lecturer in Clinical Genetics at Great Ormond Street Hospital for Children and the UCL Institute of Child Health.

Richard trained in medicine at Cambridge University and University College London. He specialized in Paediatrics and subsequently Clinical Genetics in London and completed his Ph.D. on childhood cancer syndromes at the Institute of Cancer Research. His main interests are in the clinical and molecular diagnosis of rare dysmorphic, neurological and multisystem childhood disorders. He has a particular interest in the translation of new genetic technology into clinical practice.

Marshall Summar, M.D.

Dr. Summar is Chief of the Division of Genetics and Metabolism at Children's National Health System and holds the Margaret O'Malley Professorship in Genetic Medicine. He leads the largest clinical division of its kind in the world, treating more than 7,000 patients per year with rare diseases. Dr. Summar is known for his pioneering work in caring for children diagnosed with rare diseases. Dr. Summar's laboratory works on both devices and treatments for patients with genetic and biochemical diseases and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs in Food and Drug Administration (FDA) trials for patients with congenital heart disease and premature birth. Dr. Summar serves on the Board of Directors (Treasurer) of the National Organization of Rare Diseases

(NORD) and chairs NORD's Scientific and Medical Advisory Committee. He is also the President of the Society of Inherited Metabolic Diseases and Chairs the Federal Government's Rare Disease Advisory Panel for the Patient-Centered Outcomes Institute. At NORD, Dr. Summar is spearheading an effort to develop national standards for families to build natural history registries to collect information about poorly understood rare diseases. Dr. Summar is active in newborn screening and telemedicine efforts to work in underserved areas. He joined Children's National in in 2010 from Vanderbilt University.

Durhane Wong-Rieger, Ph.D.

Dr. Wong-Rieger is President & CEO of the Canadian Organization for Rare Disorders, chair of the Council of Rare Diseases International, and chair of the Consumer Advocare Network, a national network for patient engagement in healthcare policy and advocacy. She is also President & CEO of the Institute for Optimizing Health Outcomes, providing training and direct service on health coaching and patient self-management. Dr. Wong-Rieger has served on numerous health policy advisory committees and panels and is a member of the Advisory Board for the Canadian Institutes of Health Research Institute of Genetics and the Patient Liaison Forum for the Canadian Drugs and Technologies in Health. As president of the Canadian Hemophilia Society, Dr. Wong-Rieger advocated on behalf of victims of tainted blood and was named to the Board of Directors of the Canadian Blood Services and the National Blood Safety Council. She has served on numerous health policy advisory committees and panels, including Project Coordinator for the Policy Dialogues for the Commission on the Future of Healthcare in Canada and consultant to the Ontario Premier's Advisory Board on Organ Donation. Internationally, Dr. Wong-Rieger is immediate Past-Chair of the International Alliance of Patient Organizations, Board Member representing patient interests at DIA International Association, Steering Member of the Health Technology Assessment International Patient /Citizen Involvement Interest Group and Secretary of the Asia Pacific Alliance of Rare Disease Organizations. She is Content Editor of the Patient Research Exchange and on the editorial board of The Patient. Dr. Wong-Rieger is a certified Health Coach and licensed T-Trainer with the Stanford-based Living A Healthy Life with Chronic Conditions. She has a Ph.D. in psychology from McGill University.