



Co-Chair & Members List

GLOBAL 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; Rare Diseases
International Council
Member



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

GLOBAL COMMISSION MEMBERS

The group of Global Commissioners reflects a wide range of perspectives and experiences – from patient advocacy groups, academic/research institutions, hospital and health systems, policy organizations, technology companies and biotechnology companies. Please see below for member biographies.



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



Pamela K. Gavin
Chief Strategy Officer
National Organization for Rare
Disorders



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; Rare Diseases
International Member



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





Anne O'Donnell-Luria

Associate Director, Center for Mendelian Genomics, Broad Institute; Physician, Division of Genetics and Genomics, Boston Children's Hospital



**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 Chairman, Board of Directors, National Organization for Rare Disorders



Durhane Wong-Rieger

President and Chief Executive Officer, Canadian Organization for Rare Disorders; Council Chair of Rare Diseases International



CO-CHAIR & MEMBER INFORMATION



Simon Kos, M.D.

Chief Medical Officer & Sr. Director; Microsoft Worldwide Health

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.

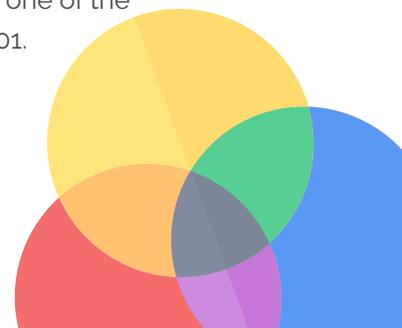


Yann Le Cam

Chief Executive Officer; EURORDIS—Rare Diseases Europe; Rare Diseases International Council Member

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.

Chief Executive Officer, Shire

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.

Chairman of Medical Genetics; King Faisal Specialist Hospital

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

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

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.



Pamela K. Gavin, MBA

Chief Strategy Officer, National Organization for Rare Disorders

Pamela Gavin sets the strategic direction for National Organization for Rare Disorders (NORD) and implements programs and services that provide innovative solutions to address the needs of the rare disease community. She is responsible for bringing together all stakeholders within the rare disease space and works closely with NORD's board of directors, donors, corporate council and member organizations, other partners, and staff. Prior to joining NORD in 2010, Pam spent 13 years executing complex, multi-stakeholder programs aimed at improving healthcare safety. As a consultant to the federal government, she implemented a new web-based portal for reporting pre-market and post-market safety data to Food and Drug Administration (FDA) and National Institutes of Health (NIH), for which she received Special Citations from the FDA Commissioner and Director of the Center for Food Safety and Applied Nutrition (CFSAN) for outstanding leadership and teamwork. Pam earned an undergraduate degree in biology from Smith College and an MBA, with a concentration in health care management, from Northeastern University.



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

Medical Genetics Service, Hospital de Clínicas de Porto Alegre; Professor of Medical Genetics, Federal University of Rio Grande do Sul, Brazil

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 he has over 450 papers published in peer-reviewed scientific journals.

**Kevin Huang**

President, Chinese Organization of Rare Disorders, Rare Diseases International Member

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 for 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 the rare disease field in China.

**Derralynn Hughes, Ph.D.**

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

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.



Anne O'Donnell-Luria, M.D., Ph.D.

Associate Director, Center for Mendelian Genomics, Broad Institute; Physician, Division of Genetics and Genomics, Boston Children's Hospital

Dr. Anne O'Donnell-Luria is a clinical and biochemical geneticist at Boston Children's Hospital and the Associate Director of the Center for Mendelian Genomics at the Broad Institute. Anne received her MD and PhD from Columbia Medical School where she studied the role of DNA methylation changes in the pathophysiology of cancer and psychiatric disease. She completed her clinical training at Boston Children's Hospital and Harvard Medical School in a combined pediatrics-genetics residency and a medical biochemical fellowship.

Anne's research in the laboratory of Daniel MacArthur focuses on improving the rate of diagnosis of rare genetic diseases. She contributes to the Genome Aggregation database (gnomAD), a variation frequency database of over 135,000 individuals from the general population, which has become a crucial part of every clinical genetics diagnosis pipeline, allowing geneticists to rule out variants with high frequencies in a variety of populations. Anne also engages in gene discovery and diagnosis in rare disease through both her position as the Associate Director for the Broad's Center for Mendelian Genomics and through her leadership role in the Rare Genomes Project, which partners directly with families to provide access to genetic research and diagnosis.

In her second life, Anne is a clinical and biochemical geneticist at Boston Children's Hospital where she runs a clinic focused on evaluating families with epigenetic disorders with rare genetic variants in genes that modify or bind histones, the proteins around which our DNA is packaged. Here she is interested in partnering with families not just for diagnosis, but also for understanding the mechanisms underlying the conditions and working towards developing epigenetic modulating therapies for these disorders.





Maryam Mohd. Fatima Matar, M.D.

Founder and Executive Director, UAE Genetic Diseases Association

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 sustainable development. She is also a globally acclaimed speaker on the topics of emerging technologies, biotechnology, epigenetics, and the impact of genetic screening and leadership.



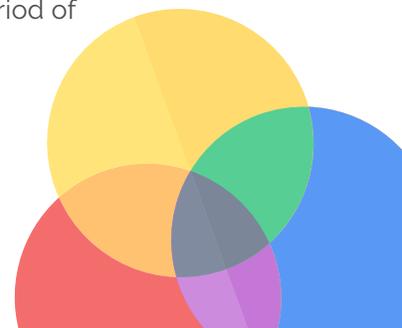
Dau-Ming Niu, M.D., Ph.D.

Director, Center for Medical Genetics, Taipei Veterans General Hospital

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

Dr. Niu received his M.D. from Kaohsiung Medical University, and he was a clinical and research fellow at the National Taiwan University Hospital. He participated in advanced studies in Professor Yuan-Tsong Chen's laboratory of 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 was 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 was the first center integrating research and medical expertise of different areas in the rare disease field in Asia.





Mike Porath

Founder and Chief Executive Officer, The Mighty

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.

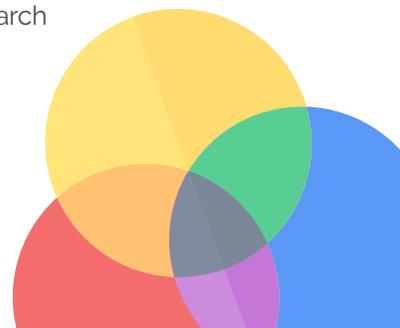
He is proud and honored to serve on this commission.



Arndt Rolfs, M.D.

Chief Executive Officer, Centogene

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. He has authored/contributed to more than 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.

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

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.

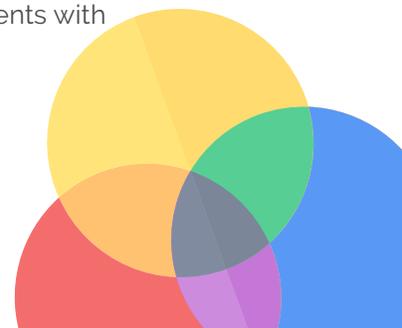
Richard trained in medicine at Cambridge University and University College London. He specialized in Paediatrics and subsequently Clinical Genetics in London and completed his Ph.D. on childhood cancer syndromes at the Institute of Cancer Research. His main interests are in the clinical and molecular 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.

Director, Rare Disease Institute; Division Chief, Genetics and Metabolism, Children's National
Chairman, Board of Directors, National Organization for Rare Disorders

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 as the Chairman of the Board of Directors at 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 2010 from Vanderbilt University.

**Durhane Wong-Rieger, Ph.D.**

President and Chief Executive Officer; Canadian Organization for Rare Disorders; Council Chair of Rare Diseases International

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.

