Rare Disease Clinical Trials Conference - Speaker Bios

Professor Elisabeth Bendstrup, Aarhus University, Denmark

Dr Bendstrup is an associate professor at the Department of Respiratory Diseases and Allergy at Aarhus University Hospital. She is responsible for the Center of Interstitial Lung Diseases, a third referral centre for interstitial lung diseases. She also takes leading part in the Danish Center of Sarcoidosis. Besides interstitial lung diseases and sarcoidosis, the center also does lung transplantation evaluation and post transplant follow-up. Dr. Bendstrup became a specialist in internal medicine in 2004 and in respiratory medicine in 2006. Dr Bendstrup's main research interest is idiopathic pulmonary fibrosis, connective-tissue disease related interstitial lung diseases, and other interstitial or rare lung diseases with specific focus on epidemiology, palliation, rehabilitation and comorbidities. She takes active part in international conferences and is member of several ILD meeting steering committees.

Professor Debra Regier, Children's National Hospital, USA

Dr. Debra Regier is the Chief for Genetics and Metabolism at Children's National Hospital. She cares for genetic, palliative care/genetic, and metabolic disease patients and families. Due to her drive to improve care for patients with Rare Diseases, she has worked to advance education across the nation through education of rare disease trainees, researchers, and primary care providers. She serves on the board of directors for the Society of Inherited Metabolic Disorders. And she is the co-director for the NORD Centers of Excellence Education committee.

Lorraine McGlinchey, PhD researcher MSc. BSc. (Hons) RGN, Ulster University

Lorraine McGlinchey Is an Adult Nurse Lecturer in the School of Nursing and Paramedic Science at Ulster University. She has a background in Orthopaedic Nursing. Lorraine is a campaigner for Sudden Adult/Arrhythmic Death syndrome and Inherited Cardiac Arrhythmias. She Is an ePAG (European Patient Advocacy Group) ERN GUARD-Heart. Her voluntary association with the Irish Heart Foundation includes membership of its Long QT support group Committee; SADs Support Group and she is a national representative on Sudden Cardiac Death Taskforce Council. Lorraine also has involvement with the Irish Inherited Cardiac Conditions Network.

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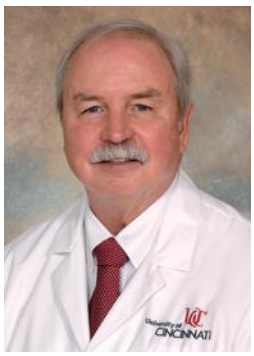
Professor Jeffrey Krischer, University of South Florida, USA

Jeffrey Krischer is Distinguished University Health Professor, Associate Chair of the Department of Internal Medicine, Morsani College of Medicine and Director of the Health Informatics Institute of the University of South Florida. He leads 200 clinical and research faculty and staff exploring the pathogenesis and prevention strategies for type 1 (autoimmune) diabetes and more than 200 rare diseases. Dr. Krischer received his Ph. D. from Harvard University. His current research focuses primarily on the design, conduct and analysis of multi-institutional clinical trials, the epidemiology of autoimmune disorders, rare diseases and cancer quality of life and symptom management.



Professor Francis X McCormack, University of Cincinnati, USA

Francis McCormack, MD, is the Gordon and Helen Hughes Taylor Chair, University of Cincinnati. He is also Professor of Medicine at the University of Cincinnati College of Medicine and the Division Director of pulmonary, critical care, and sleep medicine. His lab conducts both basic and clinical research, focused primarily on development of pathogenesis-driven molecular diagnostics and therapeutics for rare lung diseases. All basic research projects have a human clinical trial on the horizon, at least conceptually. The laboratory has used mouse models of pulmonary Langerhans cell histiocytosis, Hermansky Pudlak syndrome, lymphangioleiomyomatosis, and pulmonary alveolar microlithiasis in preclinical studies to determine mechanisms of alveolar homeostasis in health and disease. He and his team are also interested in the role of pulmonary airway cells, collectins, and lung epithelial cells in innate immune defense against inhaled bacteria, mycobacteria, fungi and viruses, especially influenza. Clinical research is focused on investigator-initiated, multicenter, international randomized trials for lymphangioleiomyomatosis, one of which recently led to discovery of an effective treatment and to FDA approval.

Professor Rachel Crowley, UCD, Dublin

Rachel Crowley is a consultant endocrinologist at SVUH and Clinical Professor at UCD. She is the co-lead (with Prof Cormac McCarthy) of the Rare Disease Clinical Trial Network. She is the co-lead (with Dr Ciara McDonnell at Children's Health Ireland) of the European Reference Network Centre for Rare Bone Disease (ERN BOND) for Irish patients. She has published on metabolic bone disease research including rare disorders and has been an investigator for academic and industry-led trials in disorders such as X-linked hypophosphataemia, osteogenesis imperfecta and familial Paget's disease. She is a clinical advisor to the Parathyroid UK patient group and the Fibrous Dysplasia Support Society UK.

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Professor Bruce Trapnell, Cincinnati Children's Hospital, USA

Dr. Trapnell is Professor of Medicine and Pediatrics at the UC College of Medicine. At Cincinnati Children's, he directed Cincinnati's Cystic Fibrosis Therapeutics Development Network Center for more than a decade and participated in numerous clinical trials as principal investigator including studies leading to FDA-approval of Creon® and Pancreaze®. He established and directs the Rare Lung Diseases Clinical Research Consortium (RLDC) in which a study led by Frank McCormack resulted in FDA approval of Sirolimus® as therapy of Lymphangioleiomyomatosis.

Dr. Trapnell's research group established that GM-CSF autoantibodies caused the rare lung disease autoimmune pulmonary alveolar proteinosis and established clinical laboratory tests for its diagnosis and identified hereditary pulmonary alveolar proteinosis as a new genetic disease caused by mutations in CSF2RA and CSF2RB. His research group developed a novel type of cell transplantation, pulmonary macrophage transplantation, and is currently translating it for testing as therapy in patients with the disease. The group also contributed to the identification, characterization, and methods for diagnosis of indium-related lung disease.

An ardent patient advocate, Dr. Trapnell has worked with patient foundations for cystic fibrosis (CF), alpha-1-antitrypsin deficiency, pulmonary alveolar proteinosis, lymphangioleiomyomatosis, and childhood interstitial lung diseases. He has served as past scientific director for the Alpha-1 Foundation and as co-founder and current scientific director of the Pulmonary Alveolar Proteinosis Foundation.

Professor David Keegan, Mater Misericordiae University Hospital Dublin

Mr Keegan's areas of special interest include inherited retinal degeneration, diabetic retinopathy, retinal transplantation (immunological consequences), health economics, low-vision device development, biomarkers in age-related macular degeneration, retinal detachment, retinal repair and regeneration. In addition to his role as consultant vitreo-retinal surgeon, Mater Hospital, Mr Keegan also serves as: National Clinical Lead for Diabetic Retinopathy, National Screening Service; Honorary Consultant Vitreo-retinal Surgeon, Temple Street Children's University Hospital; Board Member of National Council for the Blind (NCBI) and Fighting Blindness Ireland; Chair of the NCBI Vision 20/20 committee to develop a state-of-the-art low vision assistance programme in Ireland. His is involved in an active retina research group which strives to ensure that its basic sciences studies parallel those in the clinic and which has run seven clinical trials. He is a member of Scientific and Medical Advisory Board for Retina International.

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Avril Daly, CEO Retina International and President EURORDIS-Rare Disease Europe

Avril Daly is President of EURORDIS-Rare Diseases Europe and CEO of Retina International the global patient-led umbrella group dedicated to the promotion of research into Retinitis Pigmentosa (RP), Usher Syndrome, Macular Degeneration and allied retinal dystrophies. Avril previously held the role of CEO at Fighting Blindness. She is the chairperson of Rare Diseases Ireland, the Irish National Alliance for Rare Diseases, and in 2011 was appointed by the Minister of Health to the steering committee for the development of the Irish National Plan for Rare Diseases, published in 2014. Avril now sits on the oversight committee for the implementation of that plan and on the working group of the Clinical Programme for Rare Diseases, HSE. Avril is co-chair of the National Vision Coalition in Ireland. Avril was a founding board member of the Medical Research Charities Group (MRCG) and Irish Platform for Patients' Organisations, Science and Industry (IPPOSI). She was diagnosed with the rare retinal condition Retinitis Pigmentosa in 1998.

Dr Gene Sullivan, Insmed Inc

Gene joined Insmed in March 2015 as Chief Medical and Scientific Officer and in June 2017 was appointed to the newly created role of Chief Product Strategy Officer. Gene has more than 20 years of experience with a focus on pulmonary and orphan diseases. He is a medical doctor trained in internal medicine, pulmonary medicine, and critical care medicine and practiced at the Cleveland Clinic from 1995 to 1999. From 1999 through 2006, Gene held positions at the U.S. Food and Drug Administration, including Deputy Director of the Division of Pulmonary and Allergy Products. From 2007 through 2012, Gene was the Chief Medical Officer of United Therapeutics. In recent years, Gene was Vice President, Global Regulatory Affairs at AstraZeneca and consulted to companies, including Insmed, on clinical drug development and strategic regulatory matters.

Dr Laura William, St Vincent's University Hospital Dublin

Dr. Laura Williams is a neurologist from Dublin, Ireland where she earned her undergraduate medical degree and postgraduate medical doctorate in Dystonia as a Newman fellow at University College Dublin. She completed general neurology training throughout Ireland and a clinical movement disorders and deep brain stimulation fellowship at Westmead Hospital, Sydney, Australia.

Her research affiliations include the Dystonia Research Unit at St. Vincent's University Hospital, Dublin and the Movement Disorders Unit at Westmead Hospital, Sydney, where her research interests include genetics, biomarkers and endophenotypes in adult onset isolated focal dystonia. Other clinical and research interests include advanced therapies in Parkinson's disease and neurodevelopmental disorders.