Biosketch

International Conference on Clinical Research Networks for Rare Diseases

Le 253
253 Rue du Faubourg Saint-Martin
Paris, France

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Daria Julkowska

Daria Julkowska has over 15 years of experience in research and management. She is the Scientific Coordinator of the European Joint Programme on Rare Diseases that brings together different type of stakeholders (researchers, funders, clinicians & patients) from 35 countries from Europe and beyond, and also is responsible for the coordination of the IRDiRC Scientific Secretariat. This position allows her to implement the strategic rare disease research and funding recommendations of IRDiRC to the development of EJP RD which includes the participation of the European Research networks. She is involved in the rare diseases field since 2010, starting from E-Rare, the ERA-Net for Research programmes on rare diseases, where for the first two years she occupied the position of the project manager to finally (April 2013-December 2018) take over the coordination of the programme. As the coordinator, she developed and put into action a set of collaborations facilitating rare diseases research, including the partnerships with European Research Infrastructures and Patients' Organizations. She has an extensive knowledge and understanding of European funding schemes and programmes. Dr. Julkowska obtained her international PhD in molecular biology at the University of Paris XI, France and University of Gdansk, Poland in 2005. She pursued her scientific vocation by the post-doctoral experience in cellular biology, at Institut Pasteur, Paris and extensive training in communication and European Union counselling. She also holds MSc in Management of Research from the University of Paris Dauphine. In 2020 she received EURORDIS Back Pearls European Rare Diseases Leadership Award.

David Pearce

David Pearce is president of Innovation and Research for Sanford Health. He completed his Bachelor of Science with honors in biological sciences at Wolverhampton Polytechnic in 1986. He gained his doctorate in 1990 at the University of Bath, U.K., and did postdoctoral training at the University of Rochester, U.S., and Oxford University, U.K.

Dr. Pearce has been researching neuronal ceroid lipofuscinosis (Batten disease) since 1997. His research has led to the first clinical trial for Juvenile Batten disease. He has published over 100 research papers on Batten disease. He also oversees a national registry for rare diseases known as the Coordination of Rare Diseases at Sanford (CoRDS). He has served on numerous National Institutes of Health (NIH) review committees, has organized rare disease workshops for the National Institute for Neurological Disorders and Stroke (NINDS) arm of the NIH and is currently the chair of the consortium assembly for the International Rare Diseases Research Consortium (IRDiRC).

In his role as president of Innovation and Research at Sanford he is responsible for overseeing the development of research programs across Sanford's nine-state footprint, including more than 450 researchers, eight research centers and more than 300 ongoing clinical trials. He is also responsible for commercialization of select research strategies, as well as integrating Sanford Research operations into Sanford Health International Clinics. Driven by Dr. Pearce's passion for developing patient-centered, impactful research programs, Sanford Research is uniquely positioned to provide translational research that can bring important discoveries from bench to bedside.

Rima Nabbout

Rima Nabbout is Professor of Paediatric Neurology at Paris Descartes University and Director of the French Centre for Rare Epilepsies at Necker Enfants Malades, Imagine Institute (INSERM U1136), Paris, France. She received her medical degree from Saint Joseph University, Beirut, Lebanon; her paediatric board from Descartes University, Paris; and a PhD in Neurosciences from University Pierre et Marie Curie, Paris, France. She is a member of the steering committee of EPICARE (European reference network on rare and complex epilepsies), of 3 task force groups of ILAE (Nosology TF, transition TF and the regulatory affairs TF), of the EJP-RD program and of scientific committees of patient's groups on rare epilepsies. She is president of the scientific committee of the BNDMR (national bank for data on rare diseases) and member of the scientific council of INSERM. Pr Nabbout areas of research include electro clinical delineation of childhood epilepsies, guidelines on epilepsy syndromes nosology and transition from childhood into adulthood, causes, mechanisms of rare epilepsies, data mining and big data and

orphan drugs trials for rare epilepsies with development of patients' centred end points and innovative methodologies. She has authored more than 220 peer-reviewed papers and received H2020 and FP7 grants.

Tiina Urv

Tiina Urv is the program director for the Rare Diseases Clinical Research Network (RDCRN), a multidisciplinary international program in the Division of Rare Diseases Research Innovation. As the lead for the RDCRN program, Tiina collaborates with 10 NIH Institutes to manage 22 consortia and a central Data Management Coordinating Center. The RDCRN has more than 200 participating sites in 17 countries and more than 100 Patient Advocacy Groups as research partners and conducts research on about 200 rare diseases. Before joining the division, Urv was a program director in the Division of Clinical Innovation where she provided stewardship for multiple Clinical and Translational Science Awards Program hubs and worked with the Trial Innovation Network as well as NCATS' Division of Rare Diseases Research Innovation.
Urv came to the National Institutes of Health (NIH) in October 2006, working as a program director at the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) in the Intellectual and Developmental Disabilities Branch. Prior to joining NIH, she was an assistant professor at the University of Massachusetts Medical School's Eunice Kennedy Shriver Center and a research scientist at the New York State Institute for Basic Research in Developmental Disabilities. At NICHD, Urv coordinated the Hunter Kelly Newborn Screening Research Program, chaired the trans-NIH Fragile X research program, and managed a diverse portfolio of basic, behavioral and bio-behavioral research related to developmental disabilities and rare diseases.
Urv is a developmental disabilities specialist with a Ph.D. from Columbia University. She earned her undergraduate degree from the University of Washington.

Alberto Pereira

Alberto M. Pereira studied Medicine at the University of Amsterdam and specialized in internal medicine and endocrinology at the MAC in Amsterdam. His PhD degree was obtained at the University of Amsterdam and he did a postdoctoral Fellowship at the Gorlaeus Laboratory (Leiden/Amsterdam Center of Drug Research). He was appointed professor of medicine, in specific for endocrine tumors, at Leiden University per 1 nov 2012.
Alberto Pereira is the Head of the Department of Endocrinology & Metabolism at Amsterdam University Medical Centers. Prof. Pereira is the Coordinator of the European Reference Network on Rare Endocrine Conditions, and the Coordinator of the European Rare Disease Research Coordination and Support Action.

Hidehiro Mizusawa

Prof. Hidehiro Mizusawa is President Special Adviser and President Emeritus of National Center of Neurology and Psychiatry (NCNP) since April 2021. He graduated with MD in 1976 from Faculty of Medicine of Tokyo University, where he received PhD in 1983. He moved to Tsukuba University as Assistant Professor in 1984 and became Associate Professor of Department of Neurology in 1988. He has been Professor and Chairperson of Department of Neurology and Neurological Sciences, Graduate School of Medical and Dental Sciences, Tokyo Medical and Dental University (1996 - 2014), where he has been Director of Center for Brain Integration Research, Director of School of Medicine, Vice Director of the Medical Hospital and Associate Managing Trustee for Research of the University. He was Director General of NCNP Hospital since 2014 and President of NCNP since 2016. He has contributed particularly to researches on pathogeneses of ALS, Pure Akinesia/PSP, SCA, mitochondrial neuropathy, distal myopathy and Prion disease. He has been PIs of Researches on Prion disease and related subjects (2002 - 2020), PI of Initiative on Rare and Undiagnosed Diseases [IRUD] (2015 -) and PI of Integrative Research for Promotion of Rare Disease Genomic Medicine (2020 -) in Japan. He served as President of Japanese Society of Neurology (2010 - 2014) and was President of PRION 2016 in Tokyo, President of WCN 2017 in Kyoto and Vice President of ICN 2018 in Tokyo.

Matt Bolz-Johnson

Matt Bolz-Johnson joined EURORDIS in 2014 as Healthcare and Research Director, later becoming ERN & Healthcare Advisor. At EURORDIS Matt has led on advocating for the rare disease community by shaping the development and delivery of European Reference Networks (ERNs). Specifically, Matt has ensured a patient centred approach is at the heart of ERNs, through developing European Patient Advocacy Groups formally linked to each ERN.

At Rare Diseases International Matt is the Programme Director - Collaborative Global Network for Rare Diseases.

Prior to joining EURORDIS, Matt worked for 14 years in the National Health Service in England, in both hospital and commissioning trusts, where he was responsible for strategy development, clinical turnaround, quality assessment and contracting across the full spectrum of healthcare. He was a National Commissioner for ultra-rare diseases and highly specialised healthcare services. He developed national clinical networks to improve equitable access, cost efficiency and clinical effectiveness, and to promote innovation for rare cancers, genetic conditions, rare paediatric conditions, transplantations and highly specialised mental health conditions.

Marshall Summar

Dr. Summar is known for his pioneering work in caring for children diagnosed with rare diseases. He joined Children's National in 2010 from Vanderbilt University. At Children's National he led and founded the Rare Disease Institute and the Division of Genetics and Metabolism from 2010-2022, currently the largest known clinical division with over 8000 patient visits a year with rare diseases. He is the past Board Chairman for the National Organization for Rare Disorders and currently chairs their Scientific and Medical Advisory Committee. He developed the Rare Disease Institute as the first Clinical Center of Excellence designated by NORD and led the establishment of the national program NORD's Rare Disease Clinical Centers of Excellence in 2021. In 2022, he received NORD's highest honor The Lifetime Achievement Award in Rare Disease. He serves on Council at NIH, the Advisory Board of the Rare Disease Diversity Coalition, the Board of Phlow Pharmaceuticals, and others. He serves on a number of international committees and advisory bodies.

Dr. Summar's laboratory works on both devices and treatments for patients with genetic diseases and adapting knowledge from rare diseases to mainstream medicine. His work has resulted in new drugs in FDA trials for patients with congenital heart disease and sickle cell disease. This work has resulted in over 100 international patents. His laboratory is best known for its work in the rare diseases affecting nitrogen and ammonia metabolism. Dr. Summar's team has developed and shared cost-effective means to perform follow up testing for metabolic disorders in underserved countries. Dr. Summar has also organized and led a number of international work groups to develop standards of care and treatment for rare diseases resulting in significant improvements in outcomes. Dr. Summar currently is directing and developing the RareCAP system for online distribution and development of treatment protocols for rare diseases in partnership with Vanderbilt University.

Maurizio Scarpa

Prof Scarpa earned his medical degree and doctorate and completed his residency in Paediatrics at the University of Padova, Italy.

He completed a postdoctoral fellowship in molecular biology and gene expression at the European Molecular Biology Laboratory in Heidelberg (Germany), and in genetics/gene therapy at Baylor College of Medicine in Houston, Texas, USA.

Prof Scarpa has been the Director of the PhD Course on Genetics and Biochemistry at the Dept. for the Woman and Child Health at the University of Padova, Italy.

He served as vice-Dean for the International Affairs at the University of Padova and Director of the International Affairs Office at the Faculty of Medicine of the University of Padova, Italy.

Prof Scarpa has extensive expertise as a basic scientist in genetics and biotechnology, and as a clinician in the diagnosis and treatment of paediatric rare disorders, neurometabolic diseases in particular. He is especially interested in developing innovative health approaches for the diagnosis and the treatment of metabolic inherited diseases, to this aim he is also collaborating with the major Biotech Companies as external independent expert.

He presently is the Coordinator of the recently approved European Reference Network for Hereditary Metabolic Diseases, MetabERN.
Prof. Scarpa´s teaching and educational interests aim, among other, at the development of a MD/PhD European Program on Inherited Metabolic Diseases.
Prof Scarpa published about 140 international peer reviewed clinical and scientific papers, book chapters and reviews.

Hyun-Young Park

Hyun-Young Park is the director of Department of Precision Medicine at Korea National Institute of health (KNIH). Before being appointed the director, she served as the director of division of rare and cardiovascular diseases at KNIH.
Dr. Park has been trying to implement a data sharing policy in Korea for the last decade, and developed Clinical Research Information Service (CRIS), clinical research data management system (iCReaT), and Clinical and Omics Data Archive (CODA). She has also developed several nationwide clinical registries such as Korea Neonatal Registry (KNN), The Korean Organ Transplantation Registry (KOTRY), Korean acute heart failure registry (KorAHF), and Korean AMI registry (KAMIR-NIH), etc.
She received a Doctor of Medicine degree in 1990 at Yonsei University College of Medicine. She served a residency in internal medicine at Yonsei University Gangnam Hospital, and then was a Fellow in Cardiology at Severance Cardiovascular Center. And then she served as an assistant professor at Yonsei Cardiovascular Research Institute. Dr. Park joined at KNIH in 2005. She has published more than 170 articles. Her major interest in research is population genetics and cardiovascular diseases.
Now she is managing the Korean Genome and Epidemiology Study (KoGES), the Korea Biobank Project (KBP), the Korean Reference Genome (KRC) as the director of Department of Precision medicine. Now she is one of key investigators of the pilot project for 'The National Project of Bio Big Data' of Republic of Korea.

Virginie Hivert

Dr. Virginie Hivert joined EURORDIS in June 2014. Referent for EURORDIS' activities related to the development of medicines for people living with rare diseases, patient empowerment (e.g. EURORDIS Summer School) and patient engagement in medicines development.
Between June 2014 and January 2022, she has served as Observer on the EMA Committee for Orphan Medicinal Products (COMP), her role was to support the three patient representatives who are members of the COMP and also to bring an additional link between the Committee Chair, vice-Chair, Members, EMA Orphan office, and the Rare Diseases Community. During this period, she was also chairing the Therapeutic Action Group (TAG) put in place by EURORDIS to give a platform for RD patient representatives who are members of the EMA Scientific Committees (COMP, Paediatric Committee – PDCO, Committee for Advanced Therapies – CAT, Pharmacovigilance and Risk Assessment Committee – PRAC) to exchange and reflect on transversal topics in an environment where confidentiality is ensured. From March 2019 to February 2022, she has been PRAC Alternate member representing patient organizations. Since June 2022, she has been appointed as one of the Civil Society representatives on the EMA Management Board.
At global level, she is involved in the International Rare Diseases Research Consortium (IRDiRC) since its inception in 2011, first on the side of the Scientific Secretariat (prior to joining EURORDIS), then as Member of the Therapies Scientific Committee and later as its vice-Chair (March 2017 -February 2021). She is now representing EURORDIS in the IRDiRC Consortium Assembly/Patient Advocacy Constituent Committee. During these years, she has been contributing to and/or leading on several taskforces (Repurposing, Orphan Drug Development Guidebook, etc).
On top of these activities, she is involved in the development and the running of projects in which EURORDIS is partner (EUPATI, PARADIGM, c4c, etc), in the content development for EURORDIS events (ERTC, ECRD, etc) and in some of the activities of Rare Diseases International.

Birute Tumiene

Assoc.prof. Birutė Tumienė is the Head of Unit for Genetics and Genomics and a Coordinator of International Affairs in the Coordination Center for Rare Diseases at Vilnius University Hospital Santaros Klinikos. Her international rare diseases-related activities have a track record of more than 15 years, currently, she is a Vice-Chair of the Diagnostic Scientific Committee in the International Rare Diseases Research Consortium IRDiRC, a member of an operating group in the European Joint Program on Rare Diseases EJPRD, National Coordinator of Orphanet Lithuania, Advisory Committee member in the European Rare dIsease research Coordination and support Action ERICA and Lithuanian Representative in the European Reference Network Board of Member States, where she co-chairs a Working Group on ERN integration into national systems. Besides, she is a Board member of European Society of Human Genetics and one of the founders of Baltic Society of Inherited Metabolic Diseases.

Samantha Parker

Samantha is Chief Patient Access Officer at Innoskel a pioneering bioscience platform company developing transformative therapies for the unmet needs of individuals with rare bone disorders.
Samantha is an influential executive in patient advocacy and gene therapy development, who excels in the development of natural history studies, registries, novel patient-centered outcomes, early access planning and collaborative networks. She previously served as CPAO at Lysogene, and prior to that, was Director of External Affairs and Rare Disease Partnerships at Orphan Europe. Samantha was a founding member of several pilot EC European Reference Networks – virtual networks of healthcare providers aiming to facilitate discussion on complex or rare diseases. At InnoSkel she is responsible for engaging with the patient and scientific community to ensure that the voices and priorities of patients and other stakeholders are reflected in all relevant aspects of research.
Samantha is also vice-chair of the International Rare Disease Research Consortium and a Scientific Advisor to the Cure GM1 Foundation

Kyriaki Tzogani

Kyriaki Tzogani is currently a senior scientific administrator at the Orphan office in the European Medicines Agency (EMA). Kyriaki joined the EMA in 2006. She has been working at the Endocrinology office dealing with the post-authorisation procedures from 2006-2009. From 2009-2020 she has been working in the Oncology office and her main portfolios were solid tumours and haematological malignancies, including rare diseases.
Kyriaki's working experience before joining the EMA includes two years as hospital pharmacist at Papageorgiou Hospital in Thessaloniki-Greece and 2 years as Regulatory and Pharmacovigilance officer in the pharmaceutical company Roche in Athens-Greece.
Kyriaki holds a Degree in Pharmacy as well as a MSc in Industrial Pharmacy, both from the University of Athens.

Edward Neilan

Dr. Edward Neilan is the Chief Medical and Scientific Officer at the National Organization for Rare Disorders (NORD®). After completing his undergraduate degree at Yale University, Dr. Neilan earned his MD and PhD degrees from Stanford University. He then completed residency and fellowship training at Harvard Medical School, where he subsequently also served as a faculty member for more than 12 years. Dr. Neilan is triple board-certified in pediatrics, clinical genetics and clinical biochemical genetics. He is a fellow of the American Academy of Pediatrics and the American College of Medical Genetics and Genomics.
Dr. Neilan also worked on many rare disease clinical trials and has contributed to the clinical development and FDA-approval of several new rare disease therapies. Immediately prior to joining NORD, Dr. Neilan worked at Sanofi Genzyme, where he led global medical affairs for rare neurogenetic diseases and also contributed to clinical development efforts across multiple programs. Prior to that, Dr. Neilan served a term as President of the Medical Staff at Boston Children's Hospital. In addition to his full-time role at NORD, Dr. Neilan also still maintains part-time faculty appointments at Boston Children's Hospital and Harvard Medical School.

Katherine Beaverson

Katherine Beaverson is currently Executive Director, Head of Patient Advocacy and External Engagement - Rare Disease Research Unit at Pfizer. Katherine has over 17 years of experience in the biotech and pharmaceutical industry, with a focus on rare and orphan diseases. She leads the strategic planning and implementation of patient engagement activities supporting mutual areas of priority. She is both external facing, engaging with rare disease patient advocacy groups to integrate their expertise into early medicines research and development and she is also internal facing, helping to facilitate alignment among cross-functional colleagues committed to advancing Pfizer science and programs *with* rare disease patient communities. Prior to joining Pfizer Inc., she held similar positions at Boehringer Ingelheim and Amicus Therapeutics. She is professionally trained as a Genetic Counselor, having, spent 10 years at both New York Hospital-Weill Cornell Medical Center and Memorial Sloan-Kettering Cancer Center before entering industry. She received her BA from Swarthmore College and her Master of Science in Human Genetics from Sarah Lawrence College. Katherine is the current Chair of the Companies Constituent Committee of the International Rare Disease Research Consortium (IRDiRC) and a Member of the New York University Pediatric Gene Therapy Medical Ethics Working Group (PGTME).

Marisol Montolio

Marisol Montolio is Biologist and PhD in Neuroscience at the University of Barcelona. In 2006, she moved to Barcelona's Biomedical Research Park where she specialized in the identification of drugs for neurological diseases, including genetic and degenerative diseases.
Subsequently, she continued her career in rare neural pediatric diseases with genetic basis at the University of Barcelona.
Later, Dr Marisol Montolio acquired extensive experience in coordinating and executing research collaborations among academic groups, private companies and Foundations / Associations for the research of rare diseases.
Dr Marisol Montolio is the Scientific Director of Duchenne Parent Project Spain (DPPE) and Curator of the Patient Registry in Spain, promoting scientific research in Duchenne and Becker Muscular Dystrophy. Also, she is the Director of the Technological Department in DPPE.
She is also Adjunct Lecturer at the University of Barcelona, where coordinates subjects in the Master of Neurosciences of this University.
Dr Marisol Montolio has an active collaboration with EURORDIS as a member of the European Patient Advocacy Group in the ERN (Patient Advocay Board, Research Board and member of the muscle Disseases Specialist Group) and member of the Solve-RD.
Also, she is in the management committee COST Action CA17103 (Delivery of antisense RNA Therapeutics).
Dr Marisol Montolio is in the External Scientific Board of the Hospital Sant Joan de Déu, Barcelona.

Franz Schaefer

Dr. Franz Schaefer is Professor of Pediatrics and Chief of the Pediatric Nephrology Division at Heidelberg University Hospital. He received his M.D. in 1986 at Würzburg University Medical School. He performed research scholarships at the Institute of Child Health, London, the University of Virginia and Stanford University.
His research interests include the genetic basis of hereditary kidney diseases, the mechanisms and prevention of kidney disease progression, the cardiovascular, metabolic and endocrine consequences of kidney disease in children, and the mangement of end-stage kidney disease by renal replacement therapy.
He conducted numerous multicenter clinical trials and established several international research consortia including the European Study Consortium for Chronic Kidney Disease in Children (ESCAPE) and the International Pediatric Dialysis Network (IPDN). He is the current chair of the Global IPNA Registry for pediatric renal replacement therapy.
Prof. Schaefer has a special interest in rare kidney disease research. He established the PodoNet Project for Research in Steroid Resistant Nephrotic Syndrome and is a Scientific Advisory Board member of the Global aHUS Registry. Recently he founded the European Rare Kidney Disease

Reference Network (ERKNet), a consortium of 40 European Reference Centers dedicated to improve the clinical management of rare kidney disorders from diagnosis to innovative therapies. Dr. Schaefer's publication record encompasses 650 scientific articles and book chapters. With more than 27,000 citations to date, his current H-Index is 77. Dr. Schaefer also has edited several books, including the international standard textbooks 'Pediatric Kidney Disease' and 'Pediatric Dialysis'.

Mark Turner

Dr. Mark Turner is Professor of Neonatology and Research Delivery at University of Liverpool. He trained in medicine at St. Andrews and Manchester. He trained in Paediatrics in the North West of England with specialist training in Neonatal Medicine in Manchester and Liverpool.
Dr. Turner is the co-coordinator of c4c (conect4children), a large collaborative European network that aims to facilitate the development of new drugs and other therapies for the entire paediatric population.

PJ Brooks

Philip J. (P.J.) Brooks is the acting director of NCATS' Division of Rare Diseases Research Innovation. Brooks represents NCATS in the NIH-wide Gene Therapy Working Group, the Regenerative Medicine Innovation Project and the International Rare Diseases Research Consortium (IRDiRC). He also is the working group co-coordinator for the NIH Common Fund program on Somatic Cell Genome Editing, one of the leaders of the Platform Vector Gene Therapy (PaVe-GT) pilot project and the co-chair of the Bespoke Gene Therapy Consortium.
In May 2022, Brooks was selected as the recipient of the 2022 Sonia Skarlatos Public Service Award by the American Society of Gene & Cell Therapy for consistently fostering and enhancing the field of gene and cell therapy.
Brooks received his doctorate in neurobiology from The University of North Carolina at Chapel Hill. After completing a postdoctoral fellowship at The Rockefeller University, he became an investigator in the NIH intramural program, where he developed an internationally recognized research program focused on two distinct areas: the molecular basis of alcohol-related cancer, and rare neurologic diseases resulting from defective DNA repair, including xeroderma pigmentosum, Cockayne syndrome and Fanconi anemia.