

Virtual International Cystinosis Family Conference 2020

Speaker Profiles

Professor Elena Levtchenko received her MD degree from Moscow State University and a Master Degree in Medical and Pharmaceutical Research from the Free University of Brussels. She trained in Paediatrics in Brussels, followed by Paediatric Nephrology training in Nijmegen, NL. Her PhD study was performed under the leadership of Professor L Monnens, Radboud University Nijmegen. In 2011 Dr Levtchenko became Head of the Department of Paediatric Nephrology in the University Hospitals Leuven, Belgium. She is a full Professor of Pediatrics in the Katholieke Universiteit (KU) Leuven.



Professor Levtchenko's scientific work is mainly focused on unravelling pathogenesis and improving clinical care of patients with genetic renal diseases. Professor Levtchenko is Principal Investigator of a MRCG-HRB research grant co-funded by Cystinosis Ireland. She is a board member of the European Society for Paediatric Nephrology (ESPN) working group on Inherited Renal Disorders. Professor Levtchenko was elected as ESPN Secretary General in 2017.

Professor Don Cairns is Professor of Pharmaceutical and Medicinal Chemistry and Head of School at Robert Gordon University, Scotland. Professor Cairns is a member of the Royal Pharmaceutical Society (RPS), and the Association of Pharmaceutical Scientists. In 2006 he was appointed to the British Pharmacopoeia Commission and serves on an Expert Advisory Group of the Commission on Human Medicines. In 2008, Prof Cairns was appointed as a Fellow of the Royal Society of Chemistry.



Professor Cairn's research interests include the design and synthesis of selective anticancer agents, the molecular modelling of drug and DNA interactions; and the design of prodrugs for the treatment of nephropathic cystinosis.

Specific research interests include:

- The design and synthesis of novel prodrugs for the treatment of nephropathic cystinosis.
- The design and synthesis of amino-substituted anthraquinones and quinoxalines as selective ligands for higher order.

Dr Joyce Senior is Director of the Professional Doctorate in Educational Psychology. She was previously programme director for the Master of Education (Additional Support Needs) and the Postgraduate Diploma in Special Educational Needs. Her work as a primary teacher and educational psychologist traversed all sectors of the Irish education system including primary, post-primary and third level.



Her research interests focus on educational provision for children with complex behavioural and learning phenotypes associated with genetic and neurological conditions. She has published nationally and internationally in peer reviewed journals in the areas of genetic and neurological conditions (epilepsy, ADHD, Williams Syndrome, fragile x syndrome, Prader-Willi syndrome, 22q11.2 deletion syndrome) and factors influencing inclusion and belonging in school, in particular for children with additional needs and disabilities.

Dr Siobhan MacHale MB BCh BAO FRCPsych FRCPI FRCEdin MCPI

Dr Siobhan MacHale is a Consultant Liaison Psychiatrist in Beaumont Hospital and Senior Lecturer in The Royal College of Surgeons of Ireland. Following completion of medical training in Ireland, she moved to train and work as a Consultant Liaison Psychiatrist in Edinburgh, including experience in the transplant setting. She returned to work in Beaumont Hospital in 2006 as a half-time general liaison psychiatrist and half-time renal transplant psychiatrist.



Dr Rachel Bishop MD, MPH, Staff Ophthalmologist (contractor), at the National Eye Institute (NEI), National Institutes of Health, Bethesda, Maryland, USA examines and treats people who are participating in clinical trials throughout the many Institutes and Centers of the NIH. This involves monitoring for medication and treatment side-effects, managing eye conditions both related and unrelated to NIH protocols, and performing eye surgery as needed.



Since joining NEI in 2006, she has provided eye care to the many cystinosis patients who have been part of the NIH-lead cystinosis studies. She is an expert on the ocular effects of cystinosis, an orphan disease, and plays an active role nationally and internationally to help formulate disease management guidelines, further research efforts, and improve clinician and patient education.

Additional research areas include Ebola-associated eye disease and ocular graft-versus-host disease. Dr. Bishop is an NEI spokesperson and helps develop NEI's educational materials. Her additional interests include bioethics, health economics, international public health, and designing for the visually impaired. She is a member of NEI's strategic planning committee.

Professor Anuj Chauhan is the Head of Department of Chemical and Biological Engineering at the Colorado School of Mines. He was previously Professor of Chemical Engineering and Dinesh O. Shah Distinguished Faculty Fellow at the University of Florida.



Professor Chauhan received his PhD in Chemical Engineering from the University of New York and was a postdoctoral fellow with the University of California, Berkley before joining the faculty at the University of Florida in 2010.

Professor Chauhan is interested in research and development in the general area of colloids and interfacial phenomena. His current research interests include:

- Tear film dynamics
- Ophthalmic drug delivery
- Contact lenses
- Drug detoxification
- Microfluidics.

Professor Chauhan is a previous recipient of MRCG-HRB project funding with Cystinosis Ireland as a joint funding partner.

Professor Justine Bacchetta is Professor of Pediatrics, CHU de Lyon – Hôpital Femme-Mère-Enfant, Lyon, France. She specialises in pediatric nephrology and pediatric diseases of calcium and phosphate metabolism.



After a research fellowship at UCLA (Los Angeles, USA), she has four main research topics of interest: bone and mineral disorders associated to chronic kidney disease (CKD-MBD), bone disease in pediatric chronic diseases, bone disease in rare inherited renal diseases (mainly cystinosis and oxalosis), and bone physiology during growth.

She has been a council member of the European Society for Paediatric Nephrology (ESPN), and she is currently the Secretary of the French Society of Pediatric Nephrology, the Coordinator of the French Inserm/pediatrics committee, and a board-member of the CKD-MBD working group of the ESPN. She is editor for Nephrology Dialysis Transplantation (pediatric section) and Pediatric Nephrology.

She has published more than 140 publications in peer-reviewed journals.

She has given 60 invited lectures in international and national conferences, and she received in 2016 the Renee Habib award from the International Pediatric Nephrology Association (IPNA).

Christian Koepl works as physiotherapist at the Centre of Social Paediatrics in Traunstein, Kliniken Suedostbayern AG, Germany.



As specialised therapist for children with developmental disorders or handicaps, Christian attended several advanced training courses such as neurodevelopmental treatment (Bobath-Therapy).

In 2011 he completed his bachelor studies leading to a Bachelor of Arts degree in Physiotherapy at THIM University of Applied Sciences in Physiotherapy in Nieuwegein, The Netherlands.

Since 2014, Mr Koepl is member of the interdisciplinary cystinosis consultation in Germany. Together with Dr Nadine Herzig he provides the orthopaedic /physiotherapeutic consultation, during which, amongst other things, he gives individual advices to the patients for activity in everyday life, for sports as well as for training and therapy.

Improving Motor performance and capacity as a long term and sustainable process in patients with cystinosis is an enduring challenge. In this context, Mr Koepl is currently establishing and leading the Galileo-Study together with Dr Nadine Herzig and other health experts.

Dr Paul C. Grimm is a paediatric nephrologist, Palo Alto, California affiliated to the Stanford University School of Medicine.



Recruited to Stanford in 2007, he has authored ~ 90 peer-reviewed publications and is the recipient of numerous honors and awards, including listing in “The Best Doctors in America” from 2002-2020. He serves as the Medical Director of the Stanford/LPCH Pediatric Kidney Transplant Program, Medical Director of the Cystinosis Referral Clinic and Director of the Pediatric Nephrology Fellowship Program.

As Director of the Fellowship Program, Dr Grimm has conducted basic and clinical research in renal disease. Under his leadership, pediatric nephrology fellows take advantage of multiple research opportunities, including completion of a master’s degree in clinical research, or working in a basic science laboratory with an adult nephrology mentor.

Dr Grimm specialises in clinical research underpinning kidney transplantation. He is also currently the PI of a safety and effectiveness study of cysteamine in patients with cystinosis, and a Phase 2 multicenter clinical trial to evaluate the pharmacokinetics and efficacy of belatacept in transplant recipients.

He previously served as Member of the American Board of Pediatrics Subboard of Pediatric Nephrology, an Associate Editor of the Journals “Pediatric Transplantation”, and “American Journal of Transplantation”, and a Councilor of the International Pediatric Transplant Association. He is a member of the Medical Advisory Board of the Cystinosis Research Foundation.

The problem of chronic renal and hepatic injury, especially transplant rejection and scarring is the focus of Dr Grimm’s lab using techniques of computerized image analysis, immunohistochemistry, in situ hybridization and molecular biology. Studies include translational research serving as a core lab

in multi institutional clinical trials of transplant rejection and glomerulonephritis. His laboratory also looks at the origin of fibrosis in human and animal scarring models by collaboration in multicenter trials and using a rat model of kidney transplantation with subsequent analysis of the scarring process.

According to Dr Grimm:

“I met my first patient with cystinosis in 1983. He was one of the first Canadians to be treated with cysteamine. Since that time I have had the privilege to care for more than 100 Cystinosis patients and their families, mostly through the Cystinosis Referral Clinic and our ongoing research. My group was recently funded by the CRF to explore the musculoskeletal issues of patients with cystinosis using advanced imaging technologies to ultimately improve their quality of life”.

Dr Aude Servais, MD, PhD, is a senior nephrologist at the Department of Adult Nephrology and Transplantation, Necker Hospital, at Paris Descartes University, in France.



She is a referent in adult nephrology in the Reference Centre for child and adult hereditary renal diseases (MARHEA) and active members of ERKNet.

Her research interests include genetics of focal segmental glomerulonephritis, management of cystinosis in adults, and C3 glomerulonephritis. In particular, she has described the clinical presentation, outcome and genotyping of patients with juvenile and adult cystinosis and analysed the French cohort of late adolescents and adult patients with nephropathic cystinosis, showing the

impact of cysteamine therapy on the progression of the disease.

She is the Principal investigator of the ECYSCO cohort, European cystinosis cohort founded by RaDiCo, French Rare Disease Cohorts, since July 2014.

She has authored more than 50 research articles in peer-reviewed journals.

Dr Ahmed Reda is a postdoctoral fellow at KU Leuven and Vrije Universiteit Brussel, both in Belgium since 2017.



His main scientific interest is male fertility preservation. He works on unravelling the mechanism and possible fertility preservation of male cystinosis patients.

Dr Reda received his Ph.D. from Karolinska Institutet, Sweden, in 2016, with the topic “Artificial testis to study early gonadal development and male germ cell differentiation”. He has worked also as a researcher in Karolinska Institutet since 2010. Dr Reda previously graduated with an M.Sc. in molecular biology

and B.Sc. in pharmaceutical sciences.

Dr Patrick Harrison is senior lecturer in the Department of Physiology, University College, Cork. Dr Harrison leads the gene editing group for rare diseases at University College Cork in Ireland, and is the European Editor of the journal Gene Therapy.



The research focus of the Harrison Laboratory is the development of gene editing to study and potentially treat genetic disorders such as cystic fibrosis (CF), cystinosis and atopic dermatitis. Notably, Dr Harrison's research group has pioneered the use of ZFNs and Cas9/gRNA to correct the most common Cystic Fibrosis (CF) causing mutation using the homology-directed repair (HDR) pathway in cell models of disease.

The current focus of Dr Harrison's research is the optimisation of base editing techniques to repair point mutations, and NHEJ-based gene editing techniques to correct splicing and frameshift mutations directly, or by homology-independent targeted integration of superexons which can correct many different mutations within a target locus as a potential therapeutic strategy.

His research group is funded by Rare Disease Foundations in Ireland, UK and USA, and has active collaborations with researchers in London, Lisbon, Utrecht, Auckland and the USA working towards the development of gene editing as a one-time cure for rare disease.

Dr Stephanie Cherqui is Associate Professor at the Department of Pediatrics, Division of Genetics University of California, San Diego, US.



Dr Cherqui began working on cystinosis as a graduate student more than 20 years ago. Her current research focuses on the ground-breaking use of stem cell and gene therapy for multi-systemic genetic disorders and fundamental understanding of tissue repair by bone marrow stem cells.

Dr. Cherqui is the chair of the American Society of Gene and Cell Therapy (ASCGT) Gene and Cell Therapy of Genetic and Metabolic Diseases committee. She is also a member of the Scientific Review Board of the Cystinosis Research Foundation and a Scientific Council member for the

Cure Cystinosis International Registry (CCIR).

In October 2019, Jordan Janz became the first patient to receive the unique autologous stem cell treatment developed by Dr Cherqui and her team as part of a Phase I/II clinical trial which aims to test its safety and efficacy as an approach to treating cystinosis. This treatment has been developed over more than a decade of research by Dr Cherqui and her research team.

Professor Emeritus Herbie (David Richard) Newell is Professor of Drug Development at the University of Sunderland and Emeritus Professor of Cancer Therapeutics at Newcastle University, England, UK. He was Director of Translational Research at Cancer Research UK from 2006 to 2009, and the founding Scientific Director of the Northern Institute for Cancer Research at Newcastle University.



Professor Newell was involved in the development of the registered cytotoxic anticancer agents carboplatin (Paraplatin®) and raltitrexed (Tomudex®), and the first-in-class PARP inhibitor rucaparib (Rubraca®). He is an author of over 230 scientific articles and was Editor-in-Chief of the journal Cancer Chemotherapy and Pharmacology

until 2016.

In addition to his academic work, Professor Newell has consulted extensively for both pharmaceutical and biotechnology companies, and until 2016 he chaired the Medical Research Council Development Pathway Funding Scheme. In 2011, Professor Newell was elected to the UK Academy of Medical Sciences and received a C.B.E. in the 2019 New Year Honours list.

Prior to the tragic early death of Professor Roz Anderson, Professor Newell was acting as an advisor on the cystinosis project and agreed with Roz before her death that he would take over the role of Principal Investigator.

Professor Paul Goodyer is an MD, pediatric nephrologist and scientist. He is the Professor of Pediatrics and Human Genetics, McGill University in Canada.



Professor Goodyer heads a research laboratory at the McGill University Health Center Research Institute focused on molecular control of kidney development as it pertains to hereditary renal disease.

With Nicoletta Eliopoulos of the Jewish General Hospital and an international team of collaborators, he is engaged in an effort to develop stem cell therapy for cystinosis, that is particularly prevalent among French Canadians.

Professor Minnie Sarwal, M.D., Ph.D., FRCP, DCH, Professor in Residence, Surgery, Medicine and Pediatrics; Director, Kidney Pancreas Transplant; Director, Precision Transplant Medicine at UCSF; FDA Science Board member.



Professor Sarwal has over 30 years of clinical experience and 25 years of research experience in translational immunogenetics, genomics, proteomics and informatics. She holds a PhD in Molecular Genetics from Cambridge University (Christ's College), under the mentorship of Nobel Laureate Sydney Brenner; a Diploma in Child Health from London, UK; Membership of the Royal College of Physicians and elected Fellow of the Royal College of Physicians, UK.



Professor Sarwal has held the Professor of Surgery/Immunology/Peds and the Medical Director position in the Peds Kidney Transplant Program at Stanford University. She headed a personalized medicine initiative for Sutter Health and is currently Professor in Residence, Surgery, Medicine and Pediatrics at University of California, San Francisco.

Professor Sarwal's Laboratory, which has been consistently funded by the NIH, targets computational multiomics for new diagnostics and drug design for renal transplant and disease. Professor Sarwal holds awards from the Transplantation Society, Stanford University and NKF for excellence in mentorship and investigative research. She also founded and ran the Stanford Cystinosis clinic for 16 years. Professor Sarwal has also been consecutively funded by the Cystinosis foundation for over a decade. A recent development from the Sarwal Lab is a new urine test for redefinition of kidney damage and rejection, without the need of a blood test or biopsy.

Professor Sarwal is a current recipient of MRCG-HRB project funding with Cystinosis Ireland as a joint funding partner.

Dr Katharina Hohenfellner attended the Medical School at the Christian-Albrecht-Universität, Kiel, German and obtained her medical degree from the University of Kiel and of the University of Vienna, Austria.



She conducted her internship in Pediatrics at the Johannes- Gutenberg University, Mainz, Germany. Habilitation in Pediatric Nephrology.

Since 2002, Dr Hohenfellner has been an Associate Professor of Pediatrics, Johannes-Gutenberg University Mainz, Germany. She became a member of the Examination Board in Pediatric Nephrology in 2011. In 2012, she established an Interdisciplinary Cystinosis Clinic in cooperation with the German patient support group in Traunstein which continues still. In 2019, this initiative was extended and now includes the long-term implementation of the Interdisciplinary Cystinosis Clinic in Rosenheim/Bavaria.

In 2015, Dr Hohenfellner established the German Cystinosis Foundation: Major projects of the German Cystinosis Foundation include: "Save – A telemedical concept to improve clinical care and treatment in patients with Cystinosis" which is supported by the Bavarian Health Ministry (in cooperation with the Goethe-University, Frankfurt) and the Pilot-Project: "Establishing a genetic Newborn-Screening for Cystinosis and SMA in Germany", funded through the German Cystinosis Foundation, Dietmar Hopp Foundation and CRN, USA.



Dr Atif Awan is a consultant paediatric nephrologist at Temple Street Children's University Hospital with a special interest in rare kidney diseases.

Thanks to the Conference Organisers



Ms Anne Marie O'Dowd, Chairperson of Cystinosis Network Europe (CNE), Founder Chairperson of the Worldwide Cystinosis Community Advisory Board (Cystinosis CAB) and founder member of Cystinosis Ireland.

Ms O'Dowd is a founder member of Cystinosis Ireland and is the Chair of the Cystinosis Ireland Research Group. Her day job in the field of adult antenatal education where she is an antenatal teacher and tutor.

Ms O'Dowd is a long time member and the current Chairperson of Cystinosis Network Europe (CNE) which is the pan-European umbrella for cystinosis patients organisations in Europe and further afield. Ms O'Dowd has been the key driver behind a major initiative of the CNE to establish the Worldwide Cystinosis CAB under the mentorship of Eurordis. Ireland holds the chair of this group, and CNE, for the next two years. CABs, a concept developed by Eurordis, were established to enable patient groups engage meaningfully with pharmaceutical companies in relation to clinical trials and other areas. Members of the Cystinosis CAB are receiving extensive education and training in all aspects of clinical trials/treatment development so that they become patient experts. The Cystinosis CAB has expanded its remit to engage at all stages of research development, including at the early research stage, and to provide advice and the patient perspective to ensure more effective clinical trials and more efficient and speedy market access.

Ms O'Dowd is also the mother of Luke aged 18 with cystinosis who is her inspiration for all of this work.



Ms Denise Dunne is Operations Manager of Cystinosis Ireland and Secretary of CNE, Member of the Worldwide Cystinosis Advisory Board. Ms Dunne has worked with patient support and medical research charities for much of her career, including roles with the Irish Lung Fibrosis Association, Fighting Blindness, the Medical Research Charities Group and Care Alliance Ireland.

She has extensive experience in the day-to-day management of organisations and providing one-to-one support to patients and families living with life-long conditions. Her work includes building our resources and supporting our families as well as working to improve knowledge and understanding of our rare condition.

Mr Liam McFadden, Treasurer of Cystinosis Ireland and Cystinosis Network Europe

Ms Sue Maguire, Executive Member of Cystinosis Ireland

Ms Rachael Power, Executive Member of Cystinosis Ireland

Dr Ruth Davis, Research Officer, Cystinosis Ireland



Mr Mick Swift, Chairman, Cystinosis Ireland

Thank you to all of the members of the **Cystinosis Network Europe Committee** who have played a major role in determining the focus of the agenda and promoting this virtual conference with their own country members and far beyond.

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