Cystinosis Network Europe

CNE International Cystinosis Conference Leuven 2022

SPEAKERS AND PRESENTERS

Elena Levtchenko
Francesco Emma
Aurélia Bertholet-Thomas
Martine Besouw
Tjessa Bondue
David Cassiman
Ingele Casteels
Stephanie Cherqui
Anuj Chauhan
Marlies Cornelissen
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Morgan Fedorchak-DiLeo
Paul Goodyer
Valeria Graceffa
Paul Grimm
Dieter Haffner
Katharina Hohenfellner
Jennifer Hollywood
Manoe Janssen
Christian Köppl
Pascal Laforêt
Hong Liang
Graham Lipkin
Sandrine Marie
Herbie Newell
Maitena Regnier
Minnie Sarwal
Aude Servais
Rezan Topaloglu
Koonraed Veys
Lore Willem

Names are in alphabetical order. Presentation titles are in italics under each name.
(M) means: This presentation is given in the closed session for medical professionals only.
(O) means: This presenter will give his / her presentation online.

Elena Levtchenko – Conference Scientific Chair

Department of Paediatric Nephrology, University Hospitals Leuven, Belgium

Prof. Levtchenko is division chief of Pediatric Nephrology in the University Hospitals Leuven, Belgium. She is a full professor of Medicine in the Katholieke Universiteit (KU) Leuven and a group leader of the laboratory of pediatric nephrology in the same University. Dr. Levtchenko's work, described in over 250 original publications and several book chapters, is mainly focused on unraveling the disease mechanisms and improving clinical care of children with genetic kidney diseases, and in particular cytinosis. She is a chair of working group on metabolic and stone-forming of the European Reference Kidney Network.
She is a council member of the International Pediatric Nephrology Association and a chair of the professional educational committee of the European Society for Paediatric Nephrology.
Opening
Overview of Cystinosis treatment in 2022
Innovative tools for patients

Dr. Aurélia Bertholet-Thomas

Hôpital Femme Mère Enfant & Université de Lyon, France
Availability of treatment for cystinosis patients around the globe

Martine Besouw

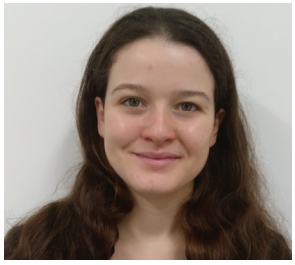
Pediatric Nephrologist, Groningen, NetherlandsL

Martine Besouw trained in Belgium (Leuven and Ghent) and the UK (London), and is now working since 5 years as a pediatric nephrologist with a special interest in tubulopathies and rare inherited kidney disorders in The Netherlands (Groningen). In 2012 she completed her PhD on cysteamine treatment for cystinosis.

Poster presentation
Future perspectives in cystinosis: From newborn screening to gene therapy (Session Chair)

Tjessa Bondue

Department of Paediatric Nephrology, University Hospitals Leuven, Belgium



Tjessa Bondue is a second year PhD student at the laboratory of Pediatric Nephrology in Leuven. She is investigating a new potential curative treatment for cystinosis, based on mRNA mediated gene replacement.

The potential of mRNA based therapy to treat nephropathic cystinosis (M)

David Cassiman

Department of hepatology and metabolic diseases, University Hospitals, Leuven, Belgium

David Cassiman is hepatologist and internist specialized on metabolic diseases. Experience: Involved in daily care for adult cystinosis patients. Involved in investigator-driven clinical research in cystinosis.

How to monitor cysteamine treatment? (Session Chair)

Ingele Casteels
Ophthalmology department at the University Hospitals, Leuven, Belgium

Ingele Casteels is the chair of the ophthalmology department at the University Hospitals Leuven. She was trained in paediatric ophthalmology by Mr David Taylor in the Great Ormond Street Hospital for Children, London and works as a paediatric ophthalmologist since 1992 in the University Hospitals Leuven. She has an interest in all fields of paediatric ophthalmology.
Panel discussion on use of eyedrops

Stephanie Cherqui
University of California, San Diego, US

Stephanie Cherqui is Associate Professor at the Department of Paediatrics, Division of Genetics University of California, San Diego, US. 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. She 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, the first patient received the unique autologous stem cell treatment developed by Stephanie 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 Stephanie Cherqui and her research team.
Status of autologous HSC transplantation in cystinosis & Condition of the patients

Anuj Chauhan
Chemical and Biological Engineering, Colorado School of Mines, Golden, US

Anuj Chauhan is Professor and Department Head in Chemical and Biological Engineering at the Colorado School of Mines. He is interested in drug delivery with a focus on treating ophthalmic diseases including cystinosis by contact lenses.
Development of contact lenses for ocular complications of cystinosis (M)

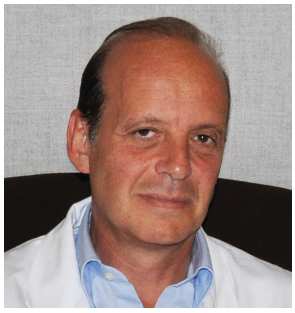
Marlies Cornelissen
Pediatric Nephrology at Radboudumc Amalia Children's Hospital, Nijmegen, Netherlands

Marlies Cornelissen is a pediatric nephrologist from Radboud umc Amalia Children's Hospital in Nijmegen, the Netherlands. She has a long standing clinical experience in rare genetic kidney diseases, like Cystinosis, in children. This clinic is the center of expertise for Cystinosis in the Netherlands.
Monitoring of cysteamine treatment patient perspective

Ester de Leo, (O)
Bambino Gesù Children's Hospital and Research Institute, Rome, Italy

Ester de Leo received her master's degree in biology in July 2006 at the University of Naples "Federico II". She worked as biologist for a few years and then she became interested in research. So, she obtained her PhD in Cellular Biochemistry in December 2011 at "Second University of Naples". Subsequently, after a first post doc in the laboratory of Prof Maria Antonietta De Matteis at the Telethon Institute of Genetics and Medicine in Naples, she moved in 2015 in the team of Prof Francesco Emma at the Bambino Gesù Children's Hospital and Research Institute in Rome. She currently collaborates on projects aimed at developing better therapies for nephropathic cystinosis. By repositioning and drug screening approach she recently identified promising compounds for cystinosis.
Genistein improves renal disease in the mouse model of nephropathic cystinosis (M)

Francesco Emma
Head of the Department of Pediatric Subspecialties and Chief of Pediatric Nephrology, Bambino Gesù Children's Hospital, Rome, Italy



Francesco Emma received his medical degree and completed his pediatric residency at the Catholic University of Louvain (Belgium). He trained in Pediatric Nephrology at Boston Children's Hospital and moved to the Bambino Gesù Children's Hospital in Rome, where he currently holds the position of Head of the Department of Pediatric Subspecialties and Chief of Pediatric Nephrology. Dr. Emma's primary research interests are in rare renal diseases, in particular in cystinosis and nephrotic syndrome. He has authored >200 research articles and several textbook chapters, and is a senior editor of the IPNA textbook "Pediatric Nephrology". He has served in several editorial boards and scientific society committees in the fields of Nephrology and Pediatric Nephrology.
Cystinosis: a global view (Session Chair)
Overview of cystinosis research

Morgan Fedorchak-DiLeo (O)
Opthalmic Biomaterials Lab, University of Pittsburgh, US

Morgan DiLeo is an Assistant Professor of Ophthalmology, Bioengineering, and Chemical Engineering at the University of Pittsburgh. As the director of the Opthalmic Biomaterials Lab, Dr. DiLeo's research interests include drug delivery to the eye and the development of novel biomaterials for ophthalmic applications.
Development and testing of a controlled release cysteamine-eyedrop (M)

John J. Foxe
Chair of the Department of Neuroscience at The University of Rochester, US

John Foxe is the Kilian J. and Caroline F. Schmitt Chair in Neuroscience Director of The Del Monte Institute for Neuroscience, and serves as Chair of the Department of Neuroscience at The University of Rochester. His research investigates the neurobiological bases of neurodevelopmental and neuropsychiatric conditions such as Autism. Dr. Foxe has authored more than 300 research and clinical papers, book chapters, commentaries, and serves as editor-in-chief of The European Journal of Neuroscience.
Neurocognitive functioning in cystinosis patients

Paul Goodyer
McGill University Health Center Research Institute, Montreal, Canada

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. He is engaged in an effort to develop stem cell therapy for cystinosis, that is particularly prevalent among French Canadians.
Prospects for translational read-through therapy of nonsense mutations in cystinosis (M)

Valeria Graceffa (O)
Biopharmaceutical Sciences, Atlantic Technological University (ATU) Sligo, Ireland

Valeria Graceffa is an Assistant Lecturer in Biopharmaceutical Sciences at the Atlantic Technological University (ATU) in Sligo, Ireland. She holds a Ph.D. in Biomedical Engineering from the National University of Ireland Galway (NUIG) and she has worked as a post-doctoral researcher at the KU Leuven University, in Belgium.
Biomaterial mediated transfer of CTNS for ocular cystinosis (M)

Paul Grimm
Stanford Children's Health Pediatric Kidney Transplant Program, Stanford, US

Paul Grimm is Professor of Pediatrics and Medical Director of the Stanford Children's Health Pediatric Kidney Transplant Program at Stanford University. He started his career as rural family doctor in Saskatchewan, Canada; then trained in Pediatrics, Nephrology and Transplantation at Winnipeg, Canada, Halifax Canada and UCLA. He had faculty appointments in Winnipeg, Canada and San Diego, California. He is Member of the Scientific Council of the Cystinosis Research Foundation.
Medications after kidney transplantation and improved treatment of infections
Stem cell transplant followed by living donor kidney transplant leaving the patient free of immunosuppression

Dieter Haffner
Department of Pediatric Kidney, Liver and Metabolic Diseases, Hannover Medical School, Hannover, Germany

Dieter Haffner is a professor of pediatrics and head of the Department of Pediatric Kidney, Liver and Metabolic Diseases and is coordinating the Center for Congenital Kidney Diseases at the Center for Rare Diseases at Hannover Medical School. His main interests are the genetic and mechanistic exploration of rare kidney diseases and mineral and bone disorders in children with chronic kidney disease (CKD-MBD). He is Assistant President of the European Society for Paediatric Nephrology (ESPN). He is a past chair of the IPNA Best Practices and Standards committee, the IPNA Publication committee, the ESPN CKD-MBD working group. He has led, or been involved in, several randomized controlled clinical trials, patient registries, and European and International Clinical Practice Guidelines (IPNA, ESPN, ERKNet).
Bone disease in cystinosis including surgery

Katharina Hohenfellner
Department for Pediatric Nephrology at RoMed Klinikum, Rosenheim, Germany

Katharina Hohenfellner is Associate Professor of Pediatrics at Johannes-Gutenberg University Mainz, Germany since 2002. In 2012 she established an Interdisciplinary Cystinosis Clinic in cooperation with the German patient support group in Traunstein. In 2019 the Interdisciplinary Cystinosis Clinic moved for a long-term implementation to Rosenheim/Bavaria. She is involved in several randomized controlled clinical trials, patient registries, and European and International Clinical Practice Guideline. The Interdisciplinary Cystinosis Clinic was accepted as a full member of ERKNet (European Reference Network for Rare Kidney Diseases) in 2022.
Cystinosis as a multi organ disease (Session Chair)
New-born screening for cystinosis & the effect of pre-symptomatic start of cysteamine treatment

Jennifer Hollywood (O)
Specialist in molecular medicine, University of Auckland, New Zealand

Jennifer Hollywood completed her PhD in molecular medicine (2014) at University College Cork (Ireland) from which she developed expertise in gene editing to model and correct genetic diseases. In 2014, she emigrated to New Zealand to continue her research in Professor Alan Davidson's lab at the University of Auckland. Here, Dr Hollywood developed the human induced pluripotent stem (iPS) cell and kidney organoid model of cystinosis. This work has shown that the iPS cell/organoid platform can be used to model aspects of cystinosis and has identified a new combination treatment. To investigate this therapy further, she has developed a rat model of cystinosis which faithfully recapitulates the human disease and her team are now performing pre-clinical drug trials in these animals. Dr Hollywood is also investigating the effects of MFSD12 knockdown on cystine levels in cystinotic iPS cells to determine whether this gene could be a therapeutic target for new cystinotic drugs.
The effects of Cysteamine and Everolimus treatment on cystinotic rats (M)
MFSD12 as a novel therapeutic target in cystinosis (M)

Manoe Janssen
Department of Pharmaceutical Sciences of the Utrecht University, Netherlands

As part of the Masereeuw group, Manoe Janssen focusses on understanding pathways that can be pharmacologically triggered to enhance the repair and regeneration of the kidney. She focusses on genetic factors and gene mutations that affect proximal tubule function and the development of human in vitro cell models for drug screening. She makes use of the unique, patented, human renal cell lines from the Masereeuw group who have a high predictive value for drug and waste product transport and metabolism. She compares cells from heathy controls and patients to understand which pathways in the cell are affected, to search for druggable targets and evaluate phenotypic improvements. As part of an international consortium on the genetic kidney disease cystinosis she is testing different drug compounds as well as CRISPR/Cas9 mediated gene repair to find a new therapy for this disease.
Advanced in vitro models for cystinosis (M)
Gene repair in cystinosis (M)

Christian Köppl
RoMed Klinikum, Rosenheim, Germany

Christian Köppl is a physiotherapist B.A. working with children with disabilities or chronic illnesses. He is a member of the interdisciplinary team at the Cystinosis Clinic in Rosenheim, Germany. With the support of the Cystinose Stiftung (Cystinosis Foundation), we are conducting the IMPACT study, a prospective study to find out whether specific muscle training with vibrating exercise equipment can improve muscle strength and endurance as well as quality of life in people with cystinosis. The study is going to end in September 2022 with the last measurements and examinations, I will report on the study protocol, the procedure and the first preliminary results.
How to improve muscle function in cystinosis: IMPACT study

Pascal Laforêt
Department of Neurology at the Versailles-Saint Quentin University, France

Pascal Laforêt, MD, PhD, is a professor of Neurology at the Versailles-Saint Quentin University, consultant specialized in neuromuscular disorders (myasthenia gravis, muscular dystrophies, and metabolic myopathies) in the Neurology department of Raymond-Poincaré hospital, and coordinator of North/East/Ile de France neuromuscular center and FHU Phenix dedicated to translational research in neuromuscular disorders. He is affiliated to U1179 INSERM-UVSQ laboratory, dedicated to biotherapies of neuromuscular system diseases. Major focus of his research activities are metabolic myopathies (pathophysiology and clinical trials), and he coordinates the French registries for mitochondrial disorders, glycogenosis type III, and Pompe disease. He is a member of the French Myology Society (SFM), French Society of Inherited Metabolic Disorders (SFEIM), and boards of the French Glycogenosis Association (AFG) and Garches Fundation.
Muscle disease in cystinosis

Hong Liang
Ophthalmologist-Praticien Hospitalier at Quinze-Vingts National Ophthalmology Hospital, Paris, France

Hong Liang is an ophthalmologist-Praticien Hospitalier at Quinze-Vingts National Ophthalmology Hospital in Paris, France. She takes care about 50 cystinosis patients in France. She conducted, as principal investigator, the Phase II/III/IV clinical trial for ocular cystinosis new treatments and is involved in the European Rare Kidney Disease Reference Network. She has 7 cystinosis related publications, and also active for cystinosis associated education and training programs.
Eye complications in cystinosis and status of eye treatment

Graham Lipkin
Department of Nephrology, University Hospital Queen Elizabeth, Edgbaston Birmingham, UK

Dr. Lipkin, BSc. MB ChB, FRCP, MD, has been clinical Director of Renal Medicine at University Hospital of Birmingham Foundation NHS Trust (UHBFT). He is Consultant Nephrologist at the Department of Renal Medicine Queen Elizabeth Hospital, University Hospitals Birmingham NHS FT & Honorary Senior Lecturer University of Birmingham He chairs the British Renal Association Clinical Affairs Committee and leads the Regional Transplant Network. His special research interest is contraception and pregnancy in women with kidney transplants

and CKD. He also does research on metabolic diseases affecting the kidney. He supervises a PhD register focused on ‘factors predictive of pre-eclampsia in women with Kidney disease‘.
Pregnancy and cystinosis

Sandrine Marie
Université Catholique de Louvain, Louvain-la-Neuve, Belgium

Sandrine Marie did her PhD in Paris, then came to Brussels in 1996. She worked for 8 years in research on purine metabolism at the de Duve Institute (with Prof van den Berghe and Prof Vincent). In 2004, I joined the metabolic diseases laboratory at Cliniques Saint-Luc, as scientific manager.

Monitoring of cysteamine treatment laboratory perspective

Herbie Newell
Drug Development at the University of Sunderland, England, UK

Herbie (David Richard) Newell is Professor of Drug Development at the University of Sunderland, England, UK, where he is continuing the work of the late Roz Anderson to develop CF10, a cysteamine prodrug for those living with cystinosis. Herbie is a cancer pharmacologist and spent 45 years discovering and developing new anticancer drugs including carboplatin, the most frequently prescribed anticancer medicine, and rucaparib, the first in patient PARP inhibitor that has remarkable activity against BRCA-related breast and ovarian cancer. Herbie is dedicated to realising the vison of Roz Anderson and creating both a lasting memorial to Roz and an effective new medicine for cystinosis.
New and improved medication for cystinosis: cysteamine prodrug

Maitena Regnier
Hôpital Femme Mère Enfant & Université de Lyon, France

Maitena Regnier is a resident in pediatrics and studying nephrology in order to become a pediatric nephrologist. She will be presenting with Dr. Aurelia Bertholet-Thomas about cystinosis around the world. She comes from Grenoble and works in Lyon this year with Dr. Bertholet-Thomas and Prof. Bachetta.

Minnie Sarwal
University of California San Francisco, US

Minnie Sarwal, MD, FRCP, DCH, PhD is Professor in Residence, Surgery, Medicine and Pediatrics, Medical Director of the Kidney Pancreas Transplant Program, Co-Director of the T32 Training Grant in transplant Surgery and Director of Precision Transplant Medicine at the University of California San Francisco (UCSF). She also holds additional positions as Adjunct Professor, Haas School of Business, University of California Berkeley, Stanford University and Adjunct Professor at the Faculty of Health Sciences, Odense, Sweden. Dr. Sarwal’s lab has been consecutively NIH funded for over 20 years and focus on computational and translational science in renal diseases, on the immunobiology and computationally driven monitoring for organ transplantation, novel the-

rapeutics and diagnostics and rational immunosuppression trial design. She has been Principal Investigator for NIH funded clinical immunosuppression trials, and is on the FDA Science Board and holds multiple positions as a Chair and Reviewer for both NIH, FDA, EU and DOD funding agencies.

Novel mechanisms and therapy for kidney injury in nephropathic cystinosis (M)

Aude Servais

Department of Adult Nephrology, at Necker Hospital, Paris, France

Aude Servais, MD, PhD, is a senior nephrologist at the Department of Adult Nephrology, at Necker Hospital, 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 in the Reference Centre for Inherited Metabolic Disorders, at Necker hospital. She is co-chair of the ERKNet working group of metabolic and stone disorders. Her research interests include the management of cystinosis in adolescents and adults.

Long-term follow-up of nephropathic cystinosis: importance of international registries

Prof. Dr. Rezan Topaloğlu

Hacettepe University Faculty of medicine Dept of Pediatric Nephrology and Rheumatology, Ankara, Turkey

Bring good treatment for cystinosis patients around the globe

Koenraad Veys

Pediatric Nephrology at the University Hospitals, Leuven, Belgium

Koenraad Veys is a paediatrician, affiliated to the Laboratory of Pediatric Nephrology of KU Leuven, who successfully defended his doctoral thesis in cystinosis entitled ‚Innovation in monitoring and treatment of nephropathic cystinosis‘ in 2019. He is currently also working as a consultant in Pediatric Nephrology at the University Hospitals Leuven, Belgium.

How to improve current treatment? (Session Chair)

Male fertility and cystinosis

Lore Willem

Psychologist, Department of Paediatric Nephrology - Organ transplantation, University Hospitals, Leuven, Belgium

Lore Willem (PhD) is a clinical child and adolescent psychologist and licensed Cognitive Behavioural Therapist. She works in the department of Paediatric Nephrology and Organ transplantation of the University Hospitals Leuven and in the Center for Anxiety and Depression, Child and Adolescent Psychiatry, University Psychiatric Center KU Leuven, Belgium.

Lore Willem is specialized in paediatric psychology and has written several articles related to this topic. In daily clinic, she offers CBT to patients with Chronic Kidney Disease and to patients with a kidney and/or liver transplant. For many years, she works with children and adolescents with Cystinosis and has offered lectures about the psychological impact of this illness on patients. She is an instructor of CBT at the University of Leuven and the University of Antwerp, Belgium and teaches Bachelor and Master’s students in psychology.

Results quality of life survey, Leuven