Cystinosis Ireland

12th Annual Dublin Cystinosis Workshop

10 February, 2026

Contents

AGENDA

PLEASE NOTE, ALL TIMES ARE IRELAND/GMT

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14:05	Prof Reza Seyedsadjadi	Harvard Medical School, Chair of Scientific Committee	Overview and Introduction
14:10	Dr Jennifer Hollywood	University College Cork, Ireland	Seedcorn scheme: Deciphering the molecular basis for how combination Cysteamine/Everolimus treatment protects against kidney damage in cystinotic rats
14:30	Dr Jennifer Hollywood	University College Cork, Ireland	HRB JFS 2022 scheme: Evaluation of a novel drug combination treatment for nephropathic cystinosis in a new cystinotic rat model
14:40	Dr Stephanie Myers	The University of Sunderland, UK	Seedcorn scheme: Development of a fluorescence-based assay for the detection of methanethiol as a cysteamine metabolite in biochemical and cellular contexts

Time	Speaker	Affiliation	Topic
15:00	Dr Francesco Bellomo	Bambino Gesù Children's Hospital, Italy	Standalone funding scheme: Generation Ctns-/- mouse on a FVB/N genetic background and comparison with C57BL/6 animals
15:10	Prof Elena Levtchenko	Amsterdam UMC, Netherlands	HRB JFS 2022 scheme: Investigating the potential of CTNS-mRNA loaded nanoparticles as a new therapeutic strategy for nephropathic cystinosis
15:30	Q&A		
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Session 2	**Chair: Dr Koenraad Veys**	**KU Leuven, Belgium**	
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Time	Speaker	Affiliation	Topic
16:50	Prof Brendan Keating	New York University - Langone Health, USA	Seedcorn Scheme: Comprehensive long-read Human Whole Genome Sequencing in >50 Kidney Transplant Recipients
17:00	Prof Anuj Chauhan	Colorado School of Mines, USA	HRB JFS 2024 scheme: Sustained delivery of cysteamine prodrugs from nanobarrier contact lenses
17:15	Dr Swastika Sur	University of California San Francisco, USA	HRB JFS 2024 scheme - Integrating Mechanistic Insights and Nano-Delivery Strategies to Treat Nephropathic Cystinosis.
17:45	Dr Tracey McCauley	Cystinosis Ireland	Overview of Cystinosis Ireland, funded projects, funding schemes
18:00	**Dr Jamie Cheh**	**Broad Institute, Massachusetts, USA**	**Keynote: Drug repurposing "The Drug repurposing Hub A resource for biological pathways Insight and Therapeutics"**
18:45	Q&A		
19:00	**Mr Mick Swift**	**Chair, Cystinosis Ireland**	**Closing remarks**
19:05 - 20:00	Catch up break out rooms		

WELCOME FROM CYSTINOSIS IRELAND

Dear Colleagues and friends,

Welcome to the 12th Annual Dublin Cystinosis Workshop. We are delighted you can be with us. Since our first Dublin Cystinosis Workshop in 2014, this meeting has grown to be the biggest annual, patient organisation-led cystinosis meeting in Europe, focussed on gathering researchers from both within and without the cystinosis research worlds to discuss areas of priority to our community. The goals of the Dublin Cystinosis Workshop are to share knowledge and ideas, build relationships and develop networks to ensure the best research is being done internationally in the field of cystinosis. I trust you will find the openness, enthusiasm and expertise of your fellow attendees bring these goals to life.

This year's meeting be a virtual (online) event owing to the CNE International Conference being hosted, in-person, by Cystinosis Ireland in Dublin in July 2026. DCW 2026 will be structured differently to previous years focusing on updates from current Cystinosis Ireland funded researchers, to showcase the breadth of research that Cystinosis Ireland fund; the ORCESTRA programme; and we will have a dedicated keynote from Dr Jaime Cheah, the Broad Institute's Drug Repurposing Hub which is an open-access, curated and annotated collection of FDA- and globally-approved drugs, clinical trial drugs, and pre-clinical tool compounds with a companion information resource. This resource is being used for unbiased exploration of pathways that affect the disease target biology, powering the discovery of new biological insights and disease mechanisms, including those in the rare disease space.

We will have presentations from seven researchers providing updates on eight projects recently funded by Cystinosis Ireland either through our Seedcorn fund, our Standalone fund or with our sister organisations and the HRB and an update from Prof Roos Masereeuw on the EU funded MSCA Joint Doctoral network ORCESTRA. Our contributors come from five countries, across Europe and North America.

This meeting is a valuable opportunity to share and discuss the cutting edge of research in rare diseases. Our focus in Cystinosis Ireland is to improve the lives of those living with cystinosis, and ultimately, to find better therapies and a cure for this condition. The Dublin Cystinosis Workshop is a vital element of that work allowing ideas to flow and grow. In bringing you together, we hope and anticipate, as has happened in the past, that the seeds of ideas will take fruit through your discussions and sharing. The open yet trusted confidence of the Dublin

Cystinosis Workshop is vital to allow freedom to create innovation. We want to work with you, both to support the development of important research questions and in competitively funding the research which aims to answer them. As it is online we will have breakout rooms organised between sessions so you will have the option to talk directly with a speaker of your choosing. We are as always immensely grateful to our Research Committee who guide and shape the agenda for the Dublin Cystinosis Workshop, identifying key themes and important speakers in and out of our world. The organising committee, chaired by Dr Reza Seyedsadjadi and supported by Anne Marie O'Dowd and Dr Tracey McCauley, have worked very hard in recent months to put the meeting together, and I thank them for this work.

We look forward to an open and engaged Dublin Cystinosis Workshop 2026.

Mick Swift
Chair, Cystinosis Ireland

Welcome from the DCW Scientific Committee

On behalf of the Scientific Organising Committee of the Dublin Cystinosis Workshop 2026, we extend a warm welcome to you at our 12th annual meeting. This workshop is a unique event in Europe – an annual gathering of a select group of global experts, each renowned in their respective fields.

A true benefit of the Dublin Cystinosis Workshop is the diversity of knowledge and expertise it brings: some attendees specialize in cystinosis research, therapies, and treatments, while others contribute innovative ideas to our shared knowledge from diverse research areas, some of which may even at first glance seem unconnected. Our years of this meeting have highlighted the value of bringing together new thinkers in different fields to enhance the understanding of all.

In other years our Workshops have covered a broad spectrum of topics related to cystinosis, but this year, we're taking a different approach. The theme for 2026 is focussed on updates from researchers who are currently funded through Cystinosis Ireland's various funding schemes. It will be a way to highlight the important and much needed research that is funded by Cystinosis and which takes place worldwide.

The Dublin Cystinosis Workshop serves as a unique platform for listening, learning, sharing, and active participation. Our lineup of speakers includes clinicians, researchers and scientists, some of whom have made, and others who are planning to make, significant contributions to the specific area, providing valuable insights and updates. Promising early career researchers and senior experts will be given the chance to showcase their work and present their ideas, underscoring the workshop's pivotal role in fostering and cultivating novel research relationships.

Collaboration is at the heart of what we will deliver in this meeting, and we aspire for attendees to leave inspired, invigorated, and prepared to tackle challenges in our collective pursuit of understanding this ultra-rare condition. This collective effort aims to pave the way for new focus areas that align closely with the needs and perspectives of people living with cystinosis.

We encourage you to join the breakout sessions to enable this collective pursuit of understanding this ultra rare disease and share new ideas and knowledge.

Warm regards,

Dr Reza Seyedsadjadi
Scientific Committee Chair

Anne Marie O'Dowd
Organising Committee Chair

WHAT IS CYSTINOSIS?

Cystinosis is a rare lysosomal storage disorder caused by autosomal recessive mutations in the CTNS gene that encodes the cystine transporter cystinosin, a ubiquitously expressed lysosomal cystine–proton co-transporter, which is expressed at the lysosomal membrane and mediates the efflux of cystine from the lysosome [1–3]. CTNS gene mutations lead to a deficiency or absence of cystinosin, with consequent accumulation of free cystine in lysosomes and buildup of toxic crystals that ultimately lead to tissue and organ damage. Cystinosis is a systemic metabolic disorder that initially affects the kidneys, as well as the eyes with accumulation of corneal cystine crystals, and, subsequently, endocrine and reproductive organs, muscles, bones, lungs, skin, and the central nervous system [4]. Based on the severity of presentation and age of onset, three clinical forms of the disease can be defined: infantile or early-onset nephropathic [5], juvenile or late-onset nephropathic [6], and the adult or ocular non-nephropathic form [7,8]. At present, more than 140 mutations have been reported [9,10], with the infantile form of cystinosis being associated with severe CTNS mutations on both alleles, and the juvenile and ocular forms mostly being associated with milder mutations in at least one allele [8,10].

The estimated incidence of cystinosis is 1 in 100,000–200,000 live births [7,8].

Snippet from the special edition of the journal, Cells, which was edited by Prof Elena Levtchenko. The above paragraph was taken from Francesco Emma, et al and Elena Levtchenko. Biomarkers in Nephropathic Cystinosis: Current and Future Perspectives. Reprinted from: Cells 2022, 11, 1839, doi:10.3390/cells11111839.

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ORGANISING COMMITTEE 2026

The Cystinosis Ireland Research Committee guides the Board in our work on identifying and supporting the best research in the field of cystinosis. The Committee was ably supported by additional experts in putting together the agenda and speakers for this meeting.

DR REZA SEYEDSADJADI

Assistant Professor of Neurology, Massachusetts General Hospital, Harvard Medical School and DCW 2026 Scientific Chair

Reza Sadjadi is an assistant professor of neurology and neuromuscular neurologist with research interests in disease outcome measures, biomarkers and clinical trial readiness in neuromuscular diseases and neuromuscular complications of rare degenerative hereditary processes. He has been involved in developing and validating multiple clinician and patient reported outcome measures using modern psychometrics and item response theory models. He is leading series of clinical trial readiness studies of distal myopathy and dysphagia in nephropathic cystinosis; clinical, neurophysiological and pathological characterization of myopathy and dysphagia in adults with nephropathic cystinosis; evaluation for inherent muscle resilience and regenerative capacity.

MS ANNE MARIE O'DOWD

Cystinosis Ireland, Cystinosis Network Europe and Worldwide Cystinosis Community Advisory Board, and DCW 2026 Organising Committee Chair

Anne Marie holds a Masters in Human Rights and Social Policy where her dissertation researched rare disease policy internationally and in Ireland. She is CEO of Alpha-1 Foundation Ireland - another rare disease - and previously worked for a human rights organisation for ethnic minorities in Ireland. She has a background in antenatal and adult education as a tutor and teacher. Anne Marie has been a board member of IPPOSI (Irish Platform for Patient Organisations) and of HRCI (Health Research Charities Ireland) and sits on the Government-appointed Rare Disease Health Technology Review Committee. Anne Marie is the mother of a young adult with cystinosis.

A founding board member of Cystinosis Ireland Anne Marie also chairs the Cystinosis Ireland Research Committee and is the chair of Cystinosis Network Europe (CNE), the umbrella group for cystinosis patient organisations in Europe and beyond, and chair of the Worldwide Cystinosis Community Advisory Board (Cystinosis CAB).

Prof Atif Awan, MD

Consultant Nephrologist, The Children's Health Ireland, Dublin Ireland

Prof Awan is a consultant paediatric nephrologist at Children's Health Ireland with a special interest in rare kidney diseases. He also is Clinical Lead at the National Rare Disease Office, HSE, Ireland.

Dr Ruth Davis

Cystinosis Ireland Research Committee

Ruth is a qualified lawyer with expertise in civil litigation, health research policy, data protection, consent for health research purposes, higher education leadership, development of strategic partnerships, advocacy with key influencers, corporate governance. Ruth is a member of the Cystinosis Ireland Research Committee. She has 20 years' experience working in both public and private sectors, particularly in the areas of research and academic management in a number of higher education institutions (HEIs) in Ireland as well in the Higher Education Authority (HEA). Ruth was appointed by the Minister for Health as a member of the National Research Ethics Committee for Medical Devices (NREC-MD).

Prof Patrick Harrison, PhD

Professor of Pediatrics, Director of Pulmonary Gene Therapy, Division of Pulmonary Medicine, Cincinnati Children's Hospital Medical Center, USA

Patrick is a Professor of Pediatrics and Director of Pulmonary Gene Therapy in the Division of Pulmonary Medicine at Cincinnati Children's Hospital Medical Center, USA. The current focus of his lab is the development of strategies to correct cystic fibrosis mutations in the CFTR gene and other rare lung disorders. Prior to moving to Cincinnati in 2024, he ran a gene editing lab at University College Cork in Ireland for two decades, and established a long term collaboration with Dr. Jennifer Hollywood in Auckland. It is purely coincidental that he moved to Cincinnati the day before Dr. Hollywood moved back to Ireland to set up her lab in Cork. Patrick helped secure the funding for the first Dublin Cystinosis workshop, and has worked with Cystinosis Ireland for many years as a co-organiser of many of the subsequent DCWs.

Eibhlis Shanahan

Community Pharmacist, Cystinosis Ireland Research Committee

Eibhlis Shanahan holds a B. Sc (Pharm), she qualified from Trinity College Dublin. She has worked as a community pharmacist for 25 years. She is involved in the training of interns to Masters level and sits on the Research Committee of Cystinosis Ireland. Most importantly, Eibhlis is the mother of a child with cystinosis.

Dr Achim Treumann

Scientific Advisor, Cystinosis Ireland Research Committee

Achim is an expert in protein chemistry and biological mass spectrometry, with an international career spanning academic core facilities, biopharma CDMOs and other biotech companies. He has led laboratories at institutions including the RCSI, the University of Newcastle and international organisations like KBI Biopharma.

Under his leadership, facilities expanded their services into recombinant protein production and advanced biophysical analysis. He has been a co-investigator on numerous international collaborations, reflected in a strong publication record. To stay at the forefront of modern science, Achim complemented his biochemistry background with an MSc in Data Science. This unique combination allows him to apply advanced data analysis and experimental design to complex biological challenges.

Until December 2025, he was the Analytical Team Lead at Paleo in Leuven, developing sustainable food. Achim has been a valued member of the Cystinosis Ireland research committee since his time in Ireland.

Dr Koenraad Veys

MD PhD, Paediatrician, Consultant in Pediatric Nephrology University Hospitals Leuven, University Hospital Ghent, Belgium, Cystinosis Ireland Research Committee

Dr Veys completed his PhD thesis in 2019 in cystinosis in the research group of Professor Elena Levtchenko and Professor Bert van den Heuvel on innovation in monitoring and treatment of nephropathic cystinosis.
His research focused on improving several contemporary aspects of the clinical management of cystinosis patients, including the development of alternative biomarkers for therapeutic monitoring, unravelling the infertility in male cystinosis patients, and developing cell-based gene therapeutic approaches to provide a cure for the kidney disease in cystinosis.

Dr Tracey McCauley

Research Manager, Cystinosis Ireland

Tracey is responsible for managing the research portfolio of the organisation, working with the Research Committee, driving and promoting the research agenda of Cystinosis Ireland.
Tracey has a background in exercise physiology, with a master's in Physical Activity and Health and a PhD in Physiology which researched Genetic Influences upon Muscle Strength and Function. Her post-doc research experience involved promoting physical activity in older people in general practice: ProAct65+ cluster randomised controlled trial. Tracey joined RCSI in 2011 where her research career evolved to project management managing an EU funded project which coordinated the surgical training of Clinical Officers in Malawi and Zambia. Upon completion of this project Tracey was assigned Research Project Manager in the UCD Diabetes Complications Research Centre. Following this post, she started her role in Cystinosis Ireland.

Denise Dunne

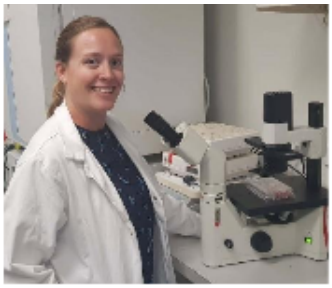
Operations Manager, Cystinosis Ireland

Denise has worked with patient support and medical research charities for much of her career, including roles with Fighting Blindness, the Medical Research Charities Group (now Health Research Charities Ireland), the Irish Lung Fibrosis Association 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.

Speaker Biographies & Abstracts

We are very much looking forward to welcoming world class researchers and clinicians to the 12th Dublin Cystinosis Workshop 2026, where we will hear the latest research updates from renowned clinicians and scientists funded by Cystinosis Ireland. We have themed this year around "Updates from current funded researchers" to showcase the breadth of research we fund, worldwide. We will have two sessions, the first starting at 14:00 (GMT) to facilitate our researchers in Europe and UK, followed by session two beginning at 16:20 (GMT) to facilitate our researchers in the US with the final presentation from our Keynote speaker Dr Jaime Cheah. We will hear from researchers and clinicians from Europe, the UK and the USA on an East to West time zone scheduling, giving their insights and most recent findings as efforts continue to identify new drug targets and effective therapies for cystinosis and ultimately to find a cure for this ultra-rare disease.

Speaker Profiles (in order of appearance)



Dr Jennifer Hollywood

Principal Investigator and Lecturer in Physiology, Department of Physiology, University College Cork, Ireland

Dr Hollywood's expertise lies in molecular medicine, gene editing technologies, and the development of disease models for therapeutic research. She works on genetic diseases such as cystic fibrosis and cystinosis, particularly in the context of developing gene therapy strategies and stem cell-based models.

She 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 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 further explore this therapeutic approach, Dr Hollywood developed a rat model of cystinosis which faithfully recapitulates the human disease. In 2024, Dr Hollywood returned to Ireland to establish her own laboratory focussed on cystinosis and kidney disease research within the Department of Physiology at UCC. Her team is currently conducting several pre-clinical drug trials using these animal models to discover and develop improved treatments for cystinosis patients.

Deciphering the molecular basis of kidney damage in cystinotic rats

Background: Cystinosis is a lysosomal storage disorder characterised by progressive renal dysfunction, with proximal tubule injury and interstitial fibrosis emerging as disease advances. While renal impairment is well established by 6 months of age in the Ctns$^{-/-}$ rat model, the molecular pathways underpinning this transition remain incompletely defined.

Methods: We performed RNA sequencing (RNA-Seq) on whole kidneys from 6-month-old Ctns$^{-/-}$ rats and age-matched wild-type controls, a time point when renal dysfunction is pronounced. Differential gene expression and pathway enrichment analyses were conducted, alongside histological assessment, to characterise transcriptional and cellular changes associated with disease progression.

Results: Cystinotic kidneys displayed coordinated suppression of renal solute transport, metabolic, and detoxification pathways. Numerous solute transporter genes were downregulated, consistent with proximal tubule dysfunction, alongside widespread reductions in amino acid, fatty acid, carbohydrate, and energy metabolism pathways, suggestive of metabolic reprogramming.

By contrast, a large number of immune-related genes were upregulated, revealing robust inflammatory activation. Increased expression of Trem2, Spp1 (osteopontin), Lgals3, C1qa/b, Vsig4, and Timd4, together with CD68^{+} and CD206^{+} macrophage infiltration, suggests the presence of TREM2/SPP1^{+} 'damage-associated' macrophages implicated in fibrotic disease. Enrichment of IL-6, IFNγ, and TNFα signalling, along with chemokines and adaptive immune markers, supports active immune trafficking and engagement of both innate and adaptive immunity. Fibrotic remodelling was evident, with upregulation of extracellular matrix genes and activation of a motile cilia programme, suggesting epithelial de-differentiation and maladaptive repair.

Conclusions: At 6 months, cystinosis induces a synchronised metabolic and transport collapse alongside immune-driven inflammation and fibrosis. Activation of TREM2/SPP1^{+} macrophages and adaptive immune pathways may help drive epithelial de-differentiation and chronic kidney disease progression, highlighting immune–fibrotic signalling as a potential therapeutic target.

Evaluation of a novel combination therapy for nephropathic cystinosis in a rat model

Background: Nephropathic cystinosis requires improved therapeutic strategies beyond standard cysteamine therapy, including better-tolerated cystine-depleting agents and adjunct treatments targeting disease mechanisms not corrected by cysteamine alone. CF10 is a novel cysteamine prodrug with improved tolerability that enables higher dosing. We have previously shown that defective autophagy in cystinotic cells is not corrected by cysteamine alone but can be restored by combination treatment with the mTOR inhibitor everolimus. Here, we evaluated whether CF10 combined with everolimus provides superior renal protection compared with CF10 monotherapy in a cystinotic rat model.

Methods: CF10 delivery and pharmacological activity were assessed relative to cysteamine using both jelly pill administration and oral gavage in male and female cystinotic rats. Long-term efficacy was first evaluated over six months using a low dose of CF10 (82.5 mg/kg) compared with cysteamine (30 mg/kg; equimolar and the maximum deliverable dose via jelly pill) to establish equivalency. A higher dose of CF10 (333 mg/kg) was then evaluated in combination with everolimus. Everolimus dosing was optimized (2 mg/kg daily) to balance efficacy and tolerability, and the CF10/everolimus combination was compared with CF10 monotherapy over six months.

Results: Both delivery routes achieved comparable cysteamine plasma levels and effectively reduced renal cystine accumulation. Low-dose CF10 performed equivalently to cysteamine in reducing tissue cystine levels, improving markers of Fanconi syndrome, and preserving renal function, confirming CF10 as an effective and better-tolerated alternative. The optimised CF10/everolimus combination demonstrated superior efficacy compared with CF10 alone, with greater cystine depletion, further improvement in Fanconi syndrome markers, and enhanced preservation of renal histology.

Conclusions: CF10 is a promising alternative to cysteamine with improved tolerability and the capacity for higher dosing. Combination therapy with everolimus provides superior renal protection compared with CF10 monotherapy, supporting a dual-targeting strategy that addresses both cystine accumulation and downstream cellular dysfunction. These findings support the translational development of CF10, which is now progressing to clinical trials.

Dr Stephanie Myers

Senior Lecturer Medicinal Chemistry, University of Sunderland, UK

Dr Myers completed her BSc (Hons) degree in Chemical and Pharmaceutical Sciences at the University of Sunderland in 2007 and undertook an industrial placement working at GlaxoSmithKline in the Psychiatry Centre of Excellence for Drug Discovery. This was followed by successful completion of a master's degree in Drug Chemistry, graduating from Newcastle University (2008). Stephanie began her research career working in the field of anti-cancer drug discovery, completing her PhD at the Northern Institute for Cancer Research, Newcastle University, in 2012 followed by a post-doctoral position between 2012-2014.

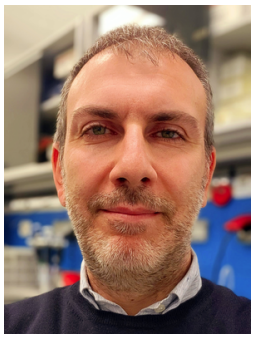
Dr Myers worked at the Institute of Cancer Research in London between 2014-2016 as a Research Fellow on the lead-optimisation of modulators of the IRE-1 enzyme and hit identification and hit-to-lead development of HSET inhibitors. She secured her first academic position at the University of Wolverhampton in 2016, beginning her independent research career as a Lecturer in Pharmaceutical Chemistry. She returned to her home city to take up a position as Senior Lecturer in Medicinal Chemistry at the University of Sunderland in 2017. Since 2019, Stephanie has worked alongside Prof Herbie Newell on the development of novel therapeutics in the rare diseases arena, following the successful identification of CF10 for the treatment of cystinosis.

Development of a fluorescence-based assay for the detection of methanethiol as a cysteamine metabolite in biochemical and cellular contexts

At the University of Sunderland, we have identified a novel cysteamine prodrug (CF10) for the treatment of cystinosis. Ongoing research in our group focuses on the development of novel cysteamine analogues with improved side effect profiles, namely, reducing or eliminating the metabolism of the drug to volatile thiol compounds (VTCs) methanethiol and dimethyl sulfide, responsible for the offensive odour associated with cysteamine therapy. As part of this drug discovery campaign, we require suitable biochemical and cellular assay methods to detect VTCs as metabolites. While the OralChroma™ system is appropriate for the detection of VTCs in oral breath, isolation of VTCs for analysis by gas chromatography presents a technical challenge for biochemical and cell-based assays.

In this project, we are investigating the use of a range of fluorescence probes from the chemical literature for their ability to react with methanethiol to give a fluorescent signal, their selectivity for methanethiol over other cellular thiols, and their suitability for incorporation into biochemical and cellular assays to detect methanethiol as a metabolic product

Dr Francesco Bellomo



Senior Researcher, Bambino Gesù Children's Hospital, Rome, Italy

Dr Francesco Bellomo is a distinguished Senior Researcher at Bambino Gesù Children's Hospital IRCCS in Rome, Italy. With a robust academic background, he earned his Master's Degree in Biological Sciences and a Ph.D. in Medical Biology and Biochemistry from the University of Bari "Aldo Moro." He further specialized in Clinical Biochemistry at the University of Rome "Tor Vergata". Dr Bellomo's research primarily focuses on nephropathic cystinosis, a rare genetic disorder. His work spans both in vitro and in vivo experiments, aiming to develop novel therapeutic strategies. He has significantly contributed to understanding the molecular and physiological mechanisms underlying cystinosis, emphasizing the importance of a multidisciplinary approach to patient care. Throughout his career, Dr Bellomo has held various academic and research positions, including a post-doctoral fellowship and a teaching role at the University of Bari. His expertise encompasses cell biology, molecular biology, and biochemistry, with skills in advanced image analysis and enzymatic assays. Dr Bellomo has been instrumental in securing substantial research funding and has published extensively in prestigious journals. His dedication to improving the quality of life for cystinosis patients is evident in his ongoing commitment to innovative research and collaboration.

Generation Ctns-/- mouse on an FVB/N genetic background and comparison with C57BL/6 animals

Mouse models remain the most widely used and validated system for studying cystinosis; however, strain and genetic background are the major determinants of the phenotype. Indeed, the Ctns−/− mice manifest delayed kidney symptoms, exhibit phenotype variability, and demonstrate strain effects, which limits their use for mechanistic and drug studies. The existence of bone phenotypes is well-documented; however, the extent of their variability is considered incomplete and is influenced by factors such as age and anatomical location. Consequently, the findings may accentuate early cellular defects while underrepresenting deformity and fragility that are evident clinically. Furthermore, certain strains have been observed to amplify inflammatory and cachexia pathways, while others have been shown to attenuate these pathways, thereby concealing systemic muscle dysfunction. In light of the aforementioned limitations, a research study was conducted in which Ctns-/- mice on the FVB/N genetic background were regenerated. These mice were originally described as a model with no proximal tubulopathy or kidney failure, but overt bone disease. To facilitate a comparison between this model and Ctns−/− mice on the C57BL/6 genetic background, both male and female cohorts will be sacrificed at 6 months of age. The objective of this study is to provide a comprehensive, multi-organ assessment of renal function, bone integrity, and muscular health.

PROF ELENA LEVTCHENKO

Full Professor, Paediatric Nephrology, Amsterdam UMC, The Netherlands

Elena Levtchenko, MD, PhD, is a full professor of pediatric nephrology, working at Emma Children Hospital in Amsterdam University Medical Centre, The Netherlands. She has devoted her clinical and research activities to improve clinical care of cystinosis patients and to unravel disease mechanisms for finding novel treatment options. She has published over 100 papers and book chapters on cystinosis and supervised several young clinicians and researchers entering cystinosis field.

Dr Levtchenko works in a tight connection with cystinosis patients organizations and chaired the working group on Inherited Metabolic Disorders of the European Reference Kidney Network (ERKNet).

mRNA Therapy for Cystinosis

For the past three decades, cystinosis has been treated with the cystine-depleting drug cysteamine. This therapy has significantly improved patient prognosis and extended life expectancy. Nevertheless, most individuals with cystinosis still develop kidney failure and extra-renal complications, albeit later in life.

The limited efficacy of cysteamine in fully curing cystinosis is believed to stem from the fact that cystinosin—the protein mutated in cystinosis—plays roles in various cellular processes beyond its primary function as a cystine transporter on the lysosomal membrane. Consequently, recent research has focused on restoring normal cystinosin protein function to address the broader spectrum of cellular dysfunction caused by its deficiency.

Our approach involves the use of CTNS messenger RNA (mRNA) as a novel form of therapy. Together with Mercurna, a Dutch biotech in mRNA-based therapeutics development, we have demonstrated the efficacy of this strategy in both cellular and zebrafish models of cystinosis. Using lipid nanoparticle (LNP)-mediated delivery, we showed dose-dependent mRNA delivery, with a detectable cystinosin expression up to 3 days. The lowest dose of CTNS mRNA-LNPs normalized cystine levels, with reduced levels persisting for at least 14 days. However, the challenge of efficiently delivering mRNA to the kidneys and other affected organs remains the focus of ongoing research. To overcome this challenge, we make use of Mercurna's targeted delivery technology for conjugating their proprietary kidney targeting peptide onto LNPs. Intravenous injection of these LNPs in wildtype mice led to delivery of CTNS mRNA in the kidney and liver.

In cystinosis PTECs, LNP transfection showed no toxicity and induced a dose-dependent cystinosin protein expression at 24 hours, remaining detectable up to 3 days post-transfection. This was associated with a dose-dependent reduction in cystine accumulation, lasting up to 14 days. In podocytes, no toxicity was observed, and we are currently evaluating cystinosin protein expression and cystine accumulation.

As the next step, targeted LNP-CTNS will be tested in the rat model of cystinosis to evaluate the expression of CTNS and its effect on cystine accumulation in the kidney.

Prof Roos Masereeuw, PhD

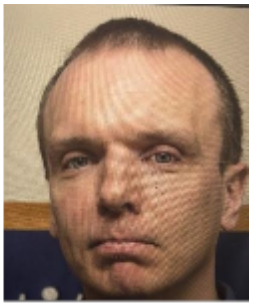
Div. Pharmacology, Utrecht Institute for Pharmaceutical Sciences, Utrecht, The Netherlands

Roos Masereeuw is full professor of Experimental Pharmacology since June 2015 at Utrecht Institute for Pharmaceutical Sciences, the Netherlands. Since February 2023, she combines her position with being the vice-Dean of Research at the Faculty of Sciences of Utrecht University. She is trained as pharmacologist and toxicologist and obtained her PhD in 1997 at Radboud University in Nijmegen, the Netherlands. Her PhD program and a postdoc period were partly performed at National Institute for Environmental Sciences (NIEHS/NIH), Research Triangle Park, North Carolina, USA. She joined the Dept. of Pharmacology and Toxicology at Radboudumc as assistant professor in 1997 and was appointed associate professor in 2002. In 2009, she received the Dutch Pharmacological Society (NVF) Schering-Plough Pharmacology Award, in 2010 the Galenus Research prize, in 2015 she was elected Fellow of the American Association of Pharmaceutical Scientists and in 2022 elected member of The Royal Holland Society of Sciences and Humanities (KHMW). She published 290 research papers and has successfully (co-)supervised 36 PhD students and currently supervises 17 PhD students, 3 assist. profs., 3 technicians and 4 post-docs.

ORGESTRA – An EU funded MSCA Joint Doctoral network

Organoids as experimental models are at the frontline of methodological development in biomedicine. These in vitro 3D cell cultures can be generated from embryonic stem cells (ESCs), induced pluripotent stem cells (iPSCs) or adult stem cells (aSCs), and can replicate organs functionally and structurally. Their physiological resemblance to target organs and ability to cryopreserve make organoids a powerful tool for biomedical research and advancing understanding of the mechanisms underlying certain disorders, including rare diseases. The ORGESTRA Joint Doctoral Network (JDN) is a multidisciplinary consortium of scientists that provides excellent training for DCs in rare disease drug development, based on innovative organoid technologies for two genetic disorders, viz. cystic fibrosis (CF) and cystinosis. Both diseases include defects in a transporter that affects epithelial tissues. For CF, the lung is highly affected while in cystinosis predominantly the kidneys are hampered. Cell-based disease models, such as organoids, capture the individual genome and in genetic diseases like CF and cystinosis, the causative factor. Effective drugs as well as robust predictive models for most rare diseases are lacking, but current developments in organoid models offer great potential as these human-based models can bridge the gap between experimental and clinical research. Importantly, many rare diseases offer high patient engagement allowing personalized model development. ORGESTRA has co-created the network around the drug development pipeline to train a new generation of scientists with all the scientific skills, and the social and management skills to become future research leaders to target unmet therapeutic needs for rare diseases through 4 integrated lines of research. This presentation will give an insight into ORGESTRA's structure and the preliminary results obtained for cystinosis with organoids.

Associate Prof Brendan Keating



Division of Transplantation, Department of Surgery, and the Institute of Systems Genetics, New York University (NYU) Langone Health, USA

Brendan Keating is an Associate Professor in the Division of Transplantation, Department of Surgery, and the Institute of Systems Genetics, New York University (NYU) Langone Health, USA, with adjunct positions in University of Pennsylvania (UPenn) and the Children's Hospital of Philadelphia (CHOP). He received his D.Phil. (Ph.D.) in molecular genetics from Christ Church College, at the University of Oxford, UK with his thesis completed in the Department of Clinical Medicine, and the Wellcome Trust Center for Human Genetics. He completed a post-doctoral fellowship at the UPenn and progressed to faculty position in the Departments of Pediatrics and Surgery. He was also a visiting Scientist at the Wellcome Trust Sanger Institute, Cambridge, UK.

His Lab research interests focus on the analyses of polymorphisms and omics transplant donor and recipient's genomes, to discover and validate genomic signals that underpin graft rejection and post-transplant complications in allotransplant, and pig to human xenotransplant settings. He instigated the formation of an international genomics consortium (iGeneTRAIN) for large-scale genomic studies using >62,000 patient DNA samples from a number of international transplant studies which is funded through various grants including an NIH-NIAID U01 award to Dr. Keating. He also leads: a Phase IV clinical trial for the early detection, diagnoses and treatment of influenza in transplant households using wearable devices (NCT06161454); and a prospective study looking at post-transplant outcomes in an NIH-NICHD funded study across 12 North American pediatric renal transplant centers (NCT03719339); and a Heart Transplant multiomics NIH funded R01. Dr. Keating also leads multiomic studies in pig to human xenotransplant studies being conducted in NYU and UPenn, and has created the largest pig to human xenotransplant biobank to date spanning sample types for up to 13 different omic studies which are funded from U19 and U01 grants.

Comprehensive long-read Human Whole Genome Sequencing in >50 Kidney Transplant Recipients

Brendan Keating [1,2,3] Kiana Moi [1,2], Francesca Zanoni [1,2], Mercy R Williams [1,2]

1. NYU Langone Transplant Institute, NYU Langone Health, New York, NY, USA
2. Department of Surgery, NYU Grossman School of Medicine, New York, NY, USA
3. Institute of Systems Genetics, NYU Langone Health, New York, NY, USA

Purpose: Whole genome sequencing (WGS) is emerging as a powerful tool for identifying clinically significant human genetic variants in transplant recipients that can influence medication selection, dosing, and immunosuppressant management. By enabling comprehensive detection of pathogenic American College of Medical Genetics and Genomics (ACMG)-'medically actionable' variants and clinically actionable pharmacogenomic (PGx) markers, WGS enhances precision medicine strategies in transplant recipients optimizing post-transplant care.

Approach: A combination of short-read and long-read WGS was performed on 52 recipient samples to identify clinically significant genetic variants. Raw sequences were aligned to the human reference genome (hg38), variants were called and annotated for functional consequences, including pathogenicity and PGx relevance. Pathogenic and likely pathogenic variants in ACMG-recommended genes were documented, while PGx variants were classified based on Clinical Pharmacogenetics Implementation Consortium (CPIC)/ and PGx Knowledge Base (PharmGKB) guidelines.

Results: ACMG analysis identified over 12 clinically significant variants across human WGS samples: including pathogenic missense variants in HFE (Hemochromatosis type 1), one likely pathogenic missense in SDHB (Pheochromocytoma), one likely pathogenic stop-gained in AMPD2 (Pontocerebellar hypoplasia type 9), and one pathogenic missense in BTD (Biotinidase deficiency). Additionally, over 40 pharmacogenes were and classified as mild, moderate, or severe based on their impact on drug dosage recommendations. Within the CYP2 gene family, we identified one poor, three rapid, and one intermediate metabolizer for CYP2C19, two intermediate metabolizers for CYP2C9, one intermediate metabolizer for CYP2D6, and one poor metabolizer for CYP2B6 which impact dosing guidelines for commonly used immunosuppressants.

Conclusions: Uncovering pathogenic variants and PGx markers provides a comprehensive genomic profile that can guide medication adjustments, immunosuppressant selection, and long-term patient monitoring. Future directions include expanding this workflow to more living transplant donors and recipients, ensuring proper return of results to enhance clinical care and advance genetics research in transplant populations.

Prof Anuj Chauhan

Chemical and Biological Engineering, Colorado School of Mines, Colorado

Anuj Chauhan received a Bachelor of Technology degree in Chemical Engineering in 1993 from Indian Institute of Technology – Delhi, India, and a PhD in Chemical Engineering in 1998 from the City University of New York.

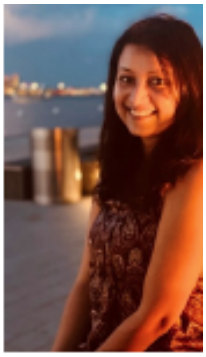
After PhD, he worked as a Post-Doctoral Researcher at the University of California at Berkeley jointly in the Departments of Chemical Engineering and Optometry. Anuj was a faculty member in the department of Chemical Engineering at the University of Florida from 2001-2018 and at the Colorado School of Mines from 2018, where he also served as the Department Head from 2018-2022. Anuj's research interests center around transport and interfacial phenomena in biological systems particularly focusing on drug delivery. He has been pursuing many problems in ophthalmology including developing novel contact lenses for improved comfort and also using contacts for delivering drugs to treat many ocular diseases including glaucoma, Cystinosis, dry eyes, infections, allergies, macular degeneration and diabetic retinopathy. He has published more than 150 publications and serves as editorial board member for The Journal of Ocular Pharmacology and Therapeutics (JOPT). Based on a recent bibliometric analysis (Bibliometric and visualized analysis of ocular drug delivery from 2001 to 2020, J. Controlled Release, 2022), he is the most productive and impactful author in ocular drug delivery.

Sustained delivery of cysteamine prodrugs from nanobarrier contact lenses

Cysteamine eye drops are utilized for treating ocular complications of cystinosis. While eye-drop based therapy is effective, it suffers from potential problems related to drug stability and compliance. We are focusing on solving this problem by designing cysteamine prodrug formulations which are stable at room temperature. The prodrugs will be converted to active cysteamine after absorption into the cornea. We have previously demonstrated that dexamethasone phosphate instilled as eyedrop is rapidly converted to dexamethasone in cornea. Based on these prior results we focus on testing feasibility of delivering cysteamine phosphate to the eye to dissolve cystine crystals in the eye. Our in vitro studies have shown that cysteamine phosphate is stable at room temperature for extended periods of time, but it converts rapidly to cysteamine in presence of alkaline phosphatase at a concentration of 10 µg/mL. We have also shown that more 1 µg of alkaline phosphatase is present in a rabbit cornea, which is about 100 µL in volume, resulting in a concentration of about 10 µg/mL. This suggests that cysteamine phosphate will readily convert to the active cysteamine in cornea. We also measured diffusion of cysteamine phosphate from control and vitamin E loaded contact lenses to explore the feasibility of sustained release by contact lenses. Future research will focus on exploring other approaches for achieving sustained release of cysteamine phosphate and measuring transport of cysteamine phosphate across a rabbit cornea ex vivo and in vivo. Additionally, we will explore other cysteamine prodrugs including thioester and carbohydrate-cysteamine thiazolidines prodrugs.

Dr Swastika Sur

University of California San Francisco, USA



Dr Swas Sur is a highly motivated and organized Molecular & Cellular Biologist with 12+ years of research experience in translational sciences. After completing her masters in Biotechnology and Ph.D. on cardiovascular diseases, in July 2017, she started her postdoctoral fellowship training, under Dr Minnie Sarwal, in cystinosis and renal transplants at UCSF.

Currently she is a Research Assistant Professor and her research interest lies in kidney diseases particularly related to kidney glomerular and tubular disorders. Dr Sur is interested in identifying and validating potential drug targets that drives inflammation and injury in kidney diseases. She has worked on –i) identifying biomarkers to differentiate types of kidney rejection post-transplant, ii) kidney tubular disorder nephropathic cystinosis, where she identified the pivotal role of V- ATPase in the disease and as a potential drug target, and iii) studied the pathogenesis of alloimmune injury and host tissue response in transplant rejection and normal human kidney.

Three of her research proposals are focused on nephropathic cystinosis and related male infertility, and identification of potential drug candidates have been funded by CRN, Cystinosis Ireland, and UCSF-Innovation Venture.

Integrating Mechanistic Insights and Nano-Delivery Strategies to Treat Nephropathic Cystinosis

Swastika Sur[1], Troy Koyama[1], Alexie Barbee[2], Bridgeen Callan[3], Tia E. Keyes[4], Jennifer Hollywood[5] and Minnie Sarwal[1]
[1] Department of Surgery, University of California San Francisco, USA
[2] Master of Translational Medicine (MTM) | UCSF/UC Berkeley
[3] School of Pharm. & Pharmaceut. Sc., Ulster University, UK
[4] School of Chemical Sciences, Dublin City University, Ireland
[5] Department of Physiology, University College Cork, Cork, Ireland

ABSTRACT

In nephropathic cystinosis, loss of cystinosin (CTNS) reduces ATP6V0A1 expression in renal proximal tubular epithelial cells (RPTECs), disrupting intracellular pH regulation, impairing mitochondrial function, and compromising autophagosome–lysosome clearance, collectively driving proximal tubular injury (Sur et al., eLife, 2024). We identified a novel oral compound, ATX, that restores ATP6V0A1 function, enhances mitochondrial–lysosomal crosstalk, and mitigates RPTEC injury. Clinical translation of ATX is limited by poor oral bioavailability. To address this, we are designing, synthesizing, and formulating drug delivery systems (DDS) capable of encapsulating ATX and related compounds, alone or in combination. In vitro and in vivo evaluation of oral nano-formulated ATX (nano-ATX) is underway.

Using a yeast split-ubiquitin two-hybrid assay, we identified 35 novel CTNS interactors. LC- MS/MS proteomic analyses revealed novel changes in cellular pathways in CTNS-/- RPTECs, both restored by ATX treatment. Leveraging intrinsic spectroscopic properties of ATX and its strong resonance Raman signature, we developed a label-free Raman imaging approach to track ATX uptake and intracellular distribution in live cells, while simultaneously monitoring anti- inflammatory effects via lipid Raman profiling. In an HCRI-HRB - Cystinosis Ireland – funded study, we are evaluating ATX, alone or with cysteamine, for improving renal Fanconi syndrome and tubular injury in a preclinical CTNS knockout rat (CTNS-/-) model. In parallel, we are developing a human kidney-on-chip platform to assess nano-ATX efficacy in a human-relevant model. Collectively, this multidisciplinary, global effort integrates mechanistic insights, target identification, and innovative drug delivery strategies to advance improved therapies for nephropathic cystinosis.

Dr Jamie Cheah

Director of Collaborative Screening, Center for the Development of Therapeutics, the Broad Institute, USA

Jaime Cheah earned her B.Sc in Biochemistry from McGill University and her Ph.D. in Neuroscience from Johns Hopkins School of Medicine in Baltimore, MD, in the laboratory of Dr. Solomon Snyder. She did her postdoctoral fellowship at the Novartis Institutes for BioMedical Research and then joined the Broad Institute as a Research Scientist, where she led a team of researchers on a large-scale cancer cell line profiling project, correlating the genetics of cancer to drug responses. She then served as the Director of the High Throughput Sciences Facility at the Koch Institute at MIT for 8 years, where she oversaw a facility outfitted with state-of-the-art equipment and helped scientists design and execute their experiments in a larger, faster and more robust way. In 2022, she returned to the Broad Institute as the Director of Collaborative Screening in the Center for the Development of Therapeutics, where she collaborates with scientists to convert their bench-top research into development of patient therapies, utilizing high throughput drug screening as part of the process. She is one of the managers of the Broad Institute's Repurposing Hub and connects with a variety of researchers, including those rare disease foundations, to enable access to this valuable resource.

The Broad Institute Repurposing Hub: a Small Molecule Toolbox for Interrogating Biological Pathways in Disease Models

The Broad Institute's Drug Repurposing Hub is an open-access, curated and annotated collection of FDA- and globally-approved drugs, clinical trial drugs, and pre-clinical tool compounds with a companion information resource. The strength of this collection is the highly-annotated resource including target, mechanisms of action, and indications for each of the ~5500 small molecules. Utilizing this collection as a "toolbox," scientists can unbiasedly uncover and explore new pathways affecting their biology of interest. In collaboration with the Center for the Development of Therapeutics at the Broad Institute, this unique collection is utilized by scientists in their high-throughput screening campaigns to identify new uses for these highly optimized small molecules in a variety of disease models. Additionally and more importantly, this resource is being used for unbiased exploration of pathways that affect the disease target biology, powering the discovery of new biological insights and disease mechanisms, including those in the rare disease space.

RESEARCH PRIORITIES

Cystinosis Ireland looks at all aspects of research that will bring us closer to improving the treatment of cystinosis, enhancing the lives of those living with the disease and taking one step closer to finding a cure.

We focus on research that aims to: (i) advance the treatment of the disease, (ii) accelerate our basic understanding of the cystinosis disease mechanisms, and (iii) enhance the quality of life for people diagnosed with cystinosis. Both basic and clinical research activities and social sciences research are of relevance to us. There are some topics of particular interest to us. These are:

Interest 1 Basic underpinning cystinosis research, including molecular and cellular biology understanding of the cystinosis disease and the development of disease models.

Interest 2 Development of new therapeutics and therapeutic targets including new drug development and stem cell therapies.

Interest 3 Understanding and tackling ocular issues in cystinosis.

Interest 4 Improving the side effects of cysteamine treatment (i.e. halitosis and body odour).

Interest 5 Understanding muscle weakness in cystinosis patients (swallowing problems have led to aspiration and death in some patients plus weakness in other muscles such as hands, legs, arms)

Interest 6 Bone weakness, including knock knees, spontaneous fractures and weak bones.

Interest 7 Not eating/vomiting particularly in early childhood. Most children need a g-tube for feeding after being diagnosed with cystinosis. A lack of eating has knock-on effects for life.

Interest 8 Developing models of care transition from childhood care to adult care in the health service.

Interest 9 Examining the social impacts of long-term childhood disease and also the impacts of treatment(s) on patient lives.

Interest 10 Psychosocial impact of living with cystinosis - is there more neurodiversity in the group vs the population?

Interest 11 Kidney Transplant and life beyond transplant

Since its establishment in 2003, Cystinosis Ireland has invested in research projects focused on all aspects of this disease to the value of €3.8 million, supporting 42 researchers, either through direct funding or as a co-funding partner with Ireland's national health research funding agency, the Health Research Board.

Cystinosis Ireland has its own Seedcorn funding scheme. The Cystinosis Ireland Seedcorn Funding Scheme aims to provide researchers with the opportunity to test new research ideas and/or to generate solid preliminary data which would contribute to a larger, sustainable, longer-term research application for funding. Typical projects considered by Cystinosis Ireland for Seedcorn funding tend to be in the region of €10,000. However, we occasionally consider applications that are for larger amounts of funding where the project is well justified and is particularly relevant to patients and families living with cystinosis.

We are particularly delighted in recent years to collaborate with sister organisations internationally in particular Cystinosis Foundation UK and Cystinosis Research Network (CRN) USA to fund excellent research. This has expanded the range and value of research and cemented our collaborative relationships with our international partners.

Since 2006, Health Research Charities Ireland (HRCI, previously MRCC) and the Health Research Board (HRB) have operated a joint funding scheme based on the dedicated allocation of funding to the HRB by the Department of Health. The HRCI/HRB joint funding scheme supports awards up to a maximum total value of €300,000 for projects from 12 up to 36 months.

This has been a very important aspect of Cystinosis Ireland's funding programme not only in providing significant funding streams but also in acknowledging world-class, cutting-edge research in the field of cystinosis. We have been very successful over the years in this highly competitive, stringently reviewed programme and were awarded two researchers' funding in the most research round of this scheme.

We would like to acknowledge and thank our international peer reviewers who work with us on this scheme and indeed, on all our research projects, without whose dedication, this work would be impossible. If you are interested in being one of our peer reviewers for upcoming research programmes, please contact us.

Please visit our website for information about applying to any of our research funding programmes. Please feel free to approach and talk with any member of our Research Committee during the DCW.

Email: **research@cystinosis.ie**
Website: **www.cystinosis.ie**

About Cystinosis Ireland

Cystinosis Ireland was founded in 2003 by those living with cystinosis and their families. The organisation continues to be led by people with personal experience of living with cystinosis. Now in our 22nd year, we are increasing the amount of work we are doing and investing in increasing numbers of research projects in areas of importance to people living with cystinosis. As of January 2026, we have funded or been a co-funding partner in research to the value of almost €4million.

Our vision

We strive for a cure for cystinosis. Until then, we want people with cystinosis to live the best life they can.

Our mission

We exist to raise awareness about cystinosis, support those affected by cystinosis, and invest in quality cystinosis research.

Our values

Empathy, determination, collaboration, innovation, integrity, professionalism.

Our Board

The Cystinosis Ireland board comprises nine individuals with an interest in cystinosis personally or professionally. The organisation's overall strategy is set by the board with four sub committees to ensure the goals are achieved: Awareness, Finance & Risk, Research and Support. Each of the groups is chaired by a board member.

The subcommittees provide an annual workplan and report on their activities at each board meeting. The subcommittees allow supporters outside the organisation to become involved on projects and activities of interest to them and where their skills and interests can be of benefit to Cystinosis Ireland.

Our Board members are:
Mick Swift, Chair
Andy Maguire, Finance & Risk subcommittee
Anne Marie O'Dowd, Chair Research subcommittee
James Ennis, Chair Support subcommittee, Northern Ireland representative
Karen McCullagh, Awareness subcommittee
Liam McFadden, Treasurer, Finance & Risk subcommittee
Rachael Power, Chair Awareness subcommittee
Sue Maguire, Support subcommittee
Tom McDonald

Our Board is supported in their work by Dr Tracey McCauley, Research Manager and Denise Dunne, Operations Manager.

Cystinosis Ireland

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