

CHAIR'S REPORT

Welcome to our Annual Report for 2024. This year has been one of continued connection, growth, and commitment to the cystinosis community.

At the heart of everything we do are people living with cystinosis, both individuals and families. To date our activities have been to support parents and families and more recently adults with cystinosis, but as our children are growing up, so are their siblings and we want to make sure we are offering helpful resources and support for every member of the family. In 2024, we made a special effort to contact adult siblings directly, hoping to build a more direct connection between them and the organisation.

Our annual Dublin Cystinosis Workshop reached its milestone 10th anniversary. The scientific organising committee again produced an excellent meeting bringing new researchers to the field and to our network together for an informative and inspiring two-day meeting. The DCW is a core part of the Cystinosis Ireland strategy to invest in and promote high quality research. The DCW has become a key annual event in the international cystinosis calendar and allows an open forum for discussion on topics of importance and relevance to the community as well as bringing experts together to develop new ideas and potential collaborations.

On the research front, we committed funding to four new projects and continued to lead in Public and Patient Involvement (PPI), helping ensure that the voices of people with cystinosis are central to scientific progress. We also supported the CNE International Conference in Manchester, where Irish and international families gathered and shared experiences.

Cystinosis Ireland launched a new website in 2024 to improve communication and accessibility, and met our fundraising target thanks to the generosity of our supporters. Our governance is strong, and we're actively planning for the future, looking at areas like transition from paediatric to adult healthcare, aging with cystinosis, newborn screening, and continued international collaboration.

Thank you to everyone who contributed to our work this year. Your time, energy, and support make a real difference. We look forward to continuing this journey with you in 2025 and beyond.

Family support

Throughout the year our Support committee continued to be a contact point for families either directly via phone and email, or through the private Facebook page. This work is led by James Ennis and Sue Maguire.

In 2024, the CNE International Conference was held in Manchester, England. As Cystinosis Ireland provides the secretariat to CNE, we provided significant operational support to the meeting through our operations manager, Denise Dunne, as well as a financial contribution. The board decided that in the years CNE hosts its international conference, instead of holding an Irish family event, Cystinosis Ireland will provide bursaries for families and people with cystinosis to travel to the CNE conference. This is to encourage families to attend a bigger meeting where more

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

connection can be built internationally. We were delighted so many attended the Manchester conference and joined us for an Irish family gathering while we were there.

Sadly, our wider family community lost a dedicated and loving supporter in 2024, with the passing of Pat Dunne, mother of one of our founders, Sue Maguire. The board offers our condolences to the Dunne and Maguire families and all who knew and loved Pat.

Research

The Cystinosis Ireland research programme continues to flourish, overseen by our research manager Dr Tracey McCauley, chair of the research committee Anne Marie O'Dowd, and the members of that committee. The work of the research committee involves identifying novel areas of research, developing relationships with researchers internationally and ensuring the research in which we invest meets the priority areas highlighted by the community.

In 2024 Cystinosis Ireland committed funding for four projects: two projects under the HRB/HRCI Joint Funding Scheme, one Seedcorn and one stand-alone. Five projects which were approved for funding in previous years continued, three jointly funded through the HRB/HRCI Joint Funding Scheme, one stand-alone, and one co-funded with Cystinosis Foundation UK.

The CI Seedcorn scheme supports projects of a shorter timeframe and more limited scope to develop data or a proof of concept to determine if a project can be viable for a larger funding scheme. Projects are typically up to six months duration and €10,000 in budget. Stand-alone project funding is awarded where a project does not fit the criteria for either of the above schemes but is deemed by the research committee to be a valuable investment in furthering our research objectives.

All research proposals recommended for funding by the research committee are reviewed by the Board for a final decision on funding.

Projects approved by the Board in 2024 were:

HRB-HRCI Joint Funding Scheme:

1. "Evaluate the protective effect of Astaxanthin for the treatment for nephropathic cystinosis in a cystinotic rat model"

Principle Investigator: Prof Minnie Sarwal, University of California, San Francisco

Amount awarded: Cystinosis Ireland total contribution €149,654 (total joint funding value: € 344,204).

2. Sustained delivery of cysteamine prodrugs from nanobarrier contact lenses

Principle Investigator: Prof Anuj Chauhan, Colorado School of Mines

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

Amount awarded: Cystinosis Ireland total contribution € 149,654. (Total joint funding value: €344,204)

Standalone Funding:

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

Principle Investigator: Dr Francesco Bellomo, Bambino Gesù Children's Hospital IRCCS, Rome

Amount awarded: €19,870.

Seedcorn Funding:

4. Deciphering the molecular basis for how combination Cysteamine/Everolimus treatment protects against kidney damage in cystinotic rats

Principle Investigators: Dr Jennifer Hollywood and Dr Alan Davidson, University of Auckland

Amount Awarded: €12,000

In other areas of importance in research, Cystinosis Ireland has become a leader in Public Patient Involvement (PPI), the process by which patient experts and advocates are involved in all stages of research design and delivery.

As part of this work, Cystinosis Ireland was confirmed as the PPI training contributor in an EU-funded PhD programme, the ORGESTRA Consortium. ORGESTRA is a Marie Skłodowska-Curie Joint Doctorate Network training 13 PhD candidates to conduct advanced research that addresses critical challenges in the field of organoid technologies for disease modelling, drug discovery and development for rare diseases namely cystic fibrosis and cystinosis. Work of this sort is essential in training future researchers and clinical investigators to understand the importance and impact that patient experts have in improving their research outcomes.

Cystinosis Ireland continues to be represented by Anne Marie O'Dowd (as chair) and James Ennis on the Worldwide Cystinosis Community Advisory Board (CAB) administered by Cystinosis Network Europe. The CAB provides an opportunity for patient advocates and experts to engage with research in all settings in a structured way.

Cystinosis Ireland was delighted in 2024 to see the return of Dr Jennifer Hollywood from the University of Auckland, New Zealand to University College Cork, to establish the first cystinosis research lab in Ireland. Dr Hollywood was funded early in her career by Cystinosis Ireland, when working with Dr Patrick Harrison in UCC, and to see her return to her alma mater to continue her work in cystinosis is a huge achievement for her and immensely valuable to the community.

Dublin Cystinosis Workshop 2024

The 10th Annual Dublin Cystinosis Workshop (DCW) took place on Friday 9th and Saturday 10th February 2024 in the Royal Marine Hotel. The meeting, which was themed Muscle and Myopathy in Cystinosis, was attended by a total of 40 people – clinicians, scientists and Cystinosis Ireland representatives. The Board took the decision to host a

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

scientific only meeting this year, to encourage family members to attend the CNE International Conference in Manchester in July.

Keynote presentations were delivered by Dr Reza Sadjadi, Harvard Medical School, USA, on Clinical trial readiness, myopathy and dysphagia assessment in adults with Nephropathic Cystinosis; and Philippos Mourikis of East-Paris University, France, on Muscle stem cells and their niche and how it can be used to study severe and mild myopathy. Both speakers were new to the DCW and to the Cystinosis Ireland network.

In total the meeting had 15 speakers (including one via Zoom) and 10 posters were presented. Video updates from currently funded researchers were provided to share their ongoing work.

We offer our congratulations to the winner of the Prof Roz Anderson Memorial Prize, Sante Princiero Berlingiero of KU Leuven, Belgium presenting his work "Targeting oxidative stress-driven lipid peroxidation improves podocyte dysfunction in cystinosis".

In acknowledgement of his extensive work with the DCW from its inception 10 years ago, and his move from University College Cork to Children's Hospital Cincinnati, Dr Patrick Harrison was presented with the gift of a wood turned bowl made by James Ennis.

Overall, the feedback from the meeting was highly positive with specific comments including:

"It was very nice and interesting to take part in the workshop. I appreciate the opportunity to learn and connect with others in the field." (F Bellomo, a new participant)

"It was a great meeting and I learned a lot" (F Emma)

"Thank you again for the wonderful meeting" (K Hohenfellner)

"Thank you so much for the kind invitation and warm hospitality. It was a true honor and pleasure to attend and connect with your team and colleagues who share similar passions." (R Sadjadi, a new participant)

"I appreciate the opportunity to engage with experts in the field and to exchange ideas on cystinosis research. The connections made and discussions held during the workshop were invaluable, and I look forward to seeing how they evolve in the future." (M Sampolesi, a new participant)

Awareness

We developed and launched a new website in 2024 – www.cystinosis.ie – to allow us to communicate better and provide better functionality and analytics. Our Awareness committee of Rachael Reilly (chair), Liam McFadden and Karen McCullagh were joined by Sinéad Maguire and Lucy Beckwith to work on developing and expanding our social media strategy and reach.

Strategy and Governance

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles. Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

In late 2024 the board began a review of our strategy to identify how it continues to meet the organisation's needs and areas where new input will be required. This process will continue in 2025.

As a regulated charity we understand the importance of being transparent in everything we do, and we aspire to continuing to be a best-in-class organisation. The board works on an ongoing basis to ensure the high level of all the work we do is reflected in our processes.

We completed our annual return to the Charity Regulator in October 2024.

Externally, our work continues to be informed by collaboration with networks and policy groups including Rare Diseases Ireland; the Irish Platform for Patient Organisations, Science and Industry; Health Research Charities Ireland; the Organ Donor Network; EURORDIS; Cystinosis Network Europe; and alliances with individual organisations with common rare disease interests.

Cystinosis Ireland continues to provide the secretariat to Cystinosis Network Europe through the work of our Operations Manager in co-ordinating CNE.

Finance

Cystinosis Ireland aims to fundraise €100,000 annually and in 2024 achieved this goal.

Funds which are donated and raised by our supporters are invested in research and family support. Ongoing costs of the organisation including staff costs and administrative overheads are met by a philanthropic donor. In 2024, our families and supporters continued to donate in generous and inventive ways including the annual Grange and Ashbourne golf days, Leixlip Tennis event, Carrigaline Ladies' Day and ongoing donations both monthly and annual. We also received a substantial corporate donation from Citco, ongoing support from Social Impact Hub in providing a registered address and meeting spaces as needed, and Cronin and Co to undertake our audit. Our supporters are many and varied and we thank each and every one for their unfailing commitment to our work. Our finance work is overseen by Andy and Sue Maguire in fundraising and Liam McFadden as Treasurer.

Staffing

Our staff at cystinosis Ireland comprises Denise Dunne, Operations Manager and Dr Tracey McCauley, Research Manager. Denise and Tracey provide our day-to-day operational support to implement the strategy as set out by the board. Their work is also key to growing our international reputation as a global leader in the cystinosis community. We encourage and welcome all members of our community to get in touch with Denise, Tracey or our board members with ideas, concerns, questions, requests or identifying support needs as they arise. We also welcome your skills and knowledge, in any way you can offer, to join our work and support the community as a whole.

Future Planning

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles. Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.



cystinosis
ireland

30 Merrion Square North
Dublin 2, Ireland
01 678 5758
www.cystinosis.ie
facebook.com/cystinosisirl
twitter.com/CystinosisIrl
Instagram.com/cystinosisireland
mail@cystinosis.ie

As people living with cystinosis grow older, the focus of our work must expand to meet their changing needs. Cystinosis Ireland is continually looking to the future and areas where we can continue to have significant impact for people with cystinosis. Areas of future focus will include newborn screening, paediatric to adult healthcare transition, emerging needs of those aging with cystinosis, and supports for siblings of people with cystinosis. We are keen to hear from anyone in the community who identifies any area where we can provide support or work meaningfully. As always, this organisation was founded and operates to offer support to those affected by cystinosis, in whatever way that might be, and we cannot do that without your input.

I would like to offer my sincere thanks to all who served on the board of Cystinosis Ireland and our committees through 2024.

I also thank everyone who has supported us in any way. This work cannot be done without you.

Mick Swift
Chair
September 2025

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

Directors: J. Ennis (NI), A. Maguire, S. Maguire, K. McCullagh, T. McDonald, L. McFadden, AM. O'Dowd, R. Reilly, M. Swift.

Patron: Stephen Rea

Charities Regulatory Authority Number: 20053796 Company Number: 371955



cystinosis
ireland

30 Merrion Square North
Dublin 2, Ireland
01 678 5758
www.cystinosis.ie
facebook.com/cystinosisirl
twitter.com/CystinosisIrl
Instagram.com/cystinosisireland
mail@cystinosis.ie

About Cystinosis Ireland

Cystinosis Ireland was founded in 2003 by people living with cystinosis and their families. The organisation continues to be led by people with personal experience of living with cystinosis.

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 Governance

Cystinosis Ireland has committed to operating to the highest standards in our governance and practice and in compliance with the Charities Governance Code. The board works continuously to implement changes as necessary and strengthen the areas where we identify weaknesses.

Our Board

In 2024, those who served on our board were: James Ennis; Andy Maguire; Sue Maguire; Karen McCullagh; Thomas McDonald; Liam McFadden; Anne Maire O'Dowd; Rachael Reilly; and Mick Swift (Chair).

The Cystinosis Ireland board comprises nine individuals with an interest in cystinosis personally or professionally.

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles. Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

Directors: J. Ennis (NI), A. Maguire, S. Maguire, K. McCullagh, T. McDonald, L. McFadden, AM. O'Dowd, R. Reilly, M. Swift.

Patron: Stephen Rea

Charities Regulatory Authority Number: 20053796 Company Number: 371955

The board is supported by our operations and research managers.

As set out in our strategic plan we have four committees – Awareness, Funding, Research and Support. Each of the committees is chaired by a board member. The committees develop an annual workplan and report on their activities at each board meeting. The committees allow supporters outside the organisation to become involved in projects and activities of interest to them and where their skills and interests can be of benefit to Cystinosis Ireland. A visual representation of organisational network is below.

A key consideration in our ongoing governance work is to identify succession planning processes. Given the very rare nature of cystinosis and the small community we represent, seeking new board members proves a challenge.

We are conscious that board members who live with cystinosis, either as an adult or a family member, tend to be long serving and bring a wealth of family, community, and first-hand experience to their role. They are experts in cystinosis. Our board members who do not live directly with the condition are experts in their own fields, including communications and research, and are committed to and engaged in our cause as friends of those living with the condition. Where new potential members of the board are identified, we hope that the committee process will allow these prospective board members to gain a deeper insight to our work and understanding of the role of the board.

Cystinosis Ireland's volunteers are primarily our board members who give of their time and expertise. The members of our committees who are not board members are also volunteers. Irish based non-board member committee members are invited to join our in-person board meetings each year as observers. This aims to give an insight into the work of the board and grow our potential pool of new board members.

We welcome and encourage members of our community and those with expertise to offer to join the team in whatever capacity is possible.

Board meeting attendance 2024

In 2024, the board met on seven occasions. Five meetings were held online, with the June and November meetings having a hybrid option – those who could not attend in person were invited to join online. At our November meeting, members of our community were invited to attend as observers, to gain a greater insight into the work of the board and the organisation. Three adults with cystinosis attended.

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

At each meeting our directors are required to declare any conflicts of interest in line with our conflict of interest policy.

The attendance at meetings is set out below.

	J Ennis	T McDonald	K McCullagh	L McFadden	AM O'Dowd	R Reilly	A Maguire	S Maguire	M Swift
January	✓	✓	✓	✓	✓	x	✓	✓	✓
February	✓	✓	✓	✓	✓	✓	✓	✓	✓
March	✓	x	✓	x	✓	x	x	✓	✓
May	✓	✓	✓	x	✓	✓	✓	✓	✓
June	✓	✓	x	✓	✓	x	x	x	✓
September	✓	x	x	x	✓	x	x	✓	✓
November	✓	✓	x	✓	✓	✓	✓	✓	✓

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.
Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.



cystinosis
ireland

30 Merrion Square North

Dublin 2, Ireland

01 678 5758

www.cystinosis.ie

facebook.com/cystinosisirl

twitter.com/CystinosisIrl

Instagram.com/cystinosisireland

mail@cystinosis.ie

Funding

Andy Maguire (Chair)

Liam McFadden

Mick Swift

Sue Maguire

Research

Anne Marie O'Dowd
(Chair)

Achim Treumann

Koenraad Veys

Eibhlís Shanahan

Ruth Davis

Tom McDonald

Tracey McCauley



Board of Directors

Mick Swift (Chair)

Andy Maguire

Anne Marie O'Dowd

James Ennis (Northern Ireland)

Karen McCullagh

Liam McFadden (Treasurer)

Rachael Reilly

Sue Maguire

Tom McDonald

Awareness

Rachael Reilly (Chair)

Liam McFadden

Karen McCullagh

Mick Swift

Sue Maguire

Support

James Ennis (Chair)

Andy Maguire

Cystinosis is
Cystinosis Ireland is a VC

Support

is a
rare condition.



cystinosis
ireland

30 Merrion Square North
Dublin 2, Ireland
01 678 5758
www.cystinosis.ie
facebook.com/cystinosisirl
twitter.com/CystinosisIrl
Instagram.com/cystinosisireland
mail@cystinosis.ie

Research - Awareness - Support

Cystinosis is a rare, degenerative, incurable disease that primarily affects children. It slowly destroys all the body's organs and muscles.

Cystinosis Ireland is a volunteer-led, non-profit organisation dedicated to funding cystinosis research and providing support to those living with the condition.

Directors: J. Ennis (NI), A. Maguire, S. Maguire, K. McCullagh, T. McDonald, L. McFadden, AM. O'Dowd, R. Reilly, M. Swift.

Patron: Stephen Rea

Charities Regulatory Authority Number: 20053796 Company Number: 371955



cystinosis
ireland

30 Merrion Square North
Dublin 2, Ireland
01 678 5758
www.cystinosis.ie
facebook.com/cystinosisirl
twitter.com/CystinosisIrl
Instagram.com/cystinosisireland
mail@cystinosis.ie

