



Chair's Report

Welcome to the Cystinosis Ireland Annual Report for 2025.

We continue to consolidate and grow while preparing for the future. Throughout 2025, Cystinosis Ireland continued to strengthen its position as a leading voice within the international cystinosis community while remaining firmly focused on our core mission of supporting people living with cystinosis and their families, investing in high-quality research and raising awareness of the condition.

One of the highlights of the year was our Dublin Cystinosis Workshop held alongside our family event. This brought together an international cohort of researchers and clinicians, with adults living with cystinosis, parents and siblings from all over Ireland. The DCW demonstrated once again the value of creating opportunities for learning, collaboration and connection.

Preparations have also been underway for the Cystinosis Network Europe International Conference, which will be held in Dublin in 2026. Following considerable discussion, planning and collaboration with partners across Europe, the Board agreed to host the conference and work has progressed rapidly to ensure an engaging and meaningful experience for families, clinicians and researchers.

Research continues to sit at the heart of our work. During 2025 we maintained funding commitments across seven active research projects, approved a new Seedcorn project and committed to participating in the next round of the HRCI-HRB Joint Funding Scheme call. We also ensured the lived experiences of people with cystinosis remain central to research design and delivery through our work in Public and Patient Involvement, and its integration into all of the research investment we make.

Our support work continued to develop during 2025. We welcomed a new family into our community, expanded resources available to families through our website, progressed work supporting adult siblings seeking genetic testing information, and continued to facilitate opportunities for adults living with cystinosis to connect with one another.

The Board also devoted significant time to strengthening governance. During the year we reviewed and adopted a number of new policies, completed a strategic review and established a Finance and Risk Committee. We continued our work to ensure full compliance with the best-practice governance standards as set out in the Governance Code.

None of this work would be possible without the commitment of our volunteers, supporters, families, researchers and professional partners. On behalf of the Board, I would like to thank everyone who has contributed their time, expertise and generosity throughout the year.

As we prepare to welcome the international cystinosis community to Dublin in 2026, we remain optimistic about the future and committed to improving the lives of those affected by cystinosis.

Research - Awareness - Support

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Patron: Stephen Rea

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Mick Swift

Chair

September 2026

Family Support

Supporting people living with cystinosis and their families remains central to everything we do.

Throughout 2025, our Support Committee, led by James Ennis and Sue Maguire, continued to provide practical and emotional support through direct contact, email, social media and family engagement activities.

The annual Dublin Cystinosis Workshop and Family Event in April brought together families from across Ireland. Attendance was strong, with new and existing families participating. The programme included medical updates, research presentations, discussions for parents, adult sessions and opportunities for social connection.

Cystinosis Ireland supports not only people living with cystinosis but also their families. In 2025, a recently diagnosed family joined our community as did the adult sibling of a person with cystinosis living overseas. We have also offered support to individuals and families internationally who do not have local supports or resources. We continue to work to ensure that families who are less connected remain informed about available supports and activities.

Progress was made in enhancing family resources, including the development of a private family section on our website which outlines various supports on offer to adults and families living in Ireland.

The year also saw continued efforts to build connections among adults living with cystinosis. Following the success of previous gatherings, adults with cystinosis continued to engage through dedicated communication channels and informal meetings. Discussions around adult healthcare services and future support needs highlighted the growing importance of services for adults living with cystinosis.

Work also progressed with Professor Willie Reardon and colleagues to provide information to adult siblings on access to genetic testing and reproductive information. Supporting siblings remains an increasingly important area of focus for the organisation.

Our committee continued to engage with rare disease and kidney disease networks in Northern Ireland and the Republic of Ireland, building relationships that may improve support options and services for our families in the future.

Research

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Research remains one of the core pillars of Cystinosis Ireland's work and is overseen by our Research Manager, Dr Tracey McCauley, working with the Research Committee chaired by Anne Marie O'Dowd, and committee members.

During 2025, Cystinosis Ireland maintained funding commitments across seven active research projects while continuing to develop new opportunities for research investment and collaboration.

Research projects

In 2025, Cystinosis Ireland continued our research investment, as outlined below. These are either ongoing or new projects, with two further grants having been approved by the Cystinosis Ireland board in the closing months of the year, to commence in 2026. You can find details of research funded by previous research on our website – www.cystinosis.ie/research

Cystinosis Ireland Seedcorn 2025

- Development of a fluorescence-based assay for the detection of methanethiol as a cysteamine metabolite in biochemical and cellular contexts – Dr Stephanie Myers, The University of Sunderland, €10,000.
- Equipment Support for the cystinosis research lab in UCC - Dr Jennifer Hollywood, University College Cork - Award Amount €3,000

HRB Joint Funding Scheme 2024

- Evaluate the protective effect of Astaxanthin for the treatment for nephropathic cystinosis in a cystinotic rat model – Prof Minnie Sarwal, UCSF, Amount awarded: CI total contribution €149,654. (Total joint funding value: € 344,204).
- Sustained delivery of cysteamine prodrugs from nanobarrier contact lenses – Prof Anuj Chauhan, Colorado School of Mines, Amount awarded: CI total contribution € 149,654. (Total joint funding value: €344,204).

Cystinosis Ireland Standalone 2024

- Generation Ctns-/- mouse on a FVB/N genetic background and comparison with C57BL/6 animals – Dr Francesco Bellomo, Bambino Gesù Children's Hospital IRCCS, €19,870.

HRB Joint Funding Scheme 2022

- Investigating the potential of CTNS-mRNA loaded nanoparticles as a new therapeutic strategy for nephropathic cystinosis - Prof Elena Levtchenko, Amsterdam University Medical Centre. Amount awarded: CI total contribution €76,200. (Total joint funding value: €152,400).

Multi-year Grants Awarded 2021

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- Clinical and Molecular Assessments of Male Infertility in Cystinosis – Prof Minnie Sarwal, UCSF, California, US – Amount Awarded: €300,000. This project is co-funded on a 50:50 basis with Cystinosis Research Network (US).

The Board also approved participation in the next HRCI-HRB Joint Funding Scheme with a commitment of up to €100,000 over three years, subject to successful applications.

Public and Patient Involvement (PPI)

Cystinosis Ireland continues to be recognised internationally as a leader in Public and Patient Involvement.

Throughout 2025, Dr Tracey McCauley and Anne Marie O'Dowd continued their involvement with the ORGESTRA Marie Skłodowska-Curie Doctoral Network, delivering training and workshops focused on integrating patient perspectives into research design and delivery. In March, they contributed to a training week with a session entitled *Patient roles in biomedical research – How to include Public Patient Involvement in Research (PPI). Using Patient organisations for PPI aspects.*

Cystinosis Ireland also continued to participate in the Worldwide Cystinosis Community Advisory Board (CAB), ensuring patient voices remain represented in research and industry discussions.

Dublin Cystinosis Workshop 2025

The Dublin Cystinosis Workshop and Family Event took place in April 2025, with the theme *Kidney aspects of cystinosis - emphasising research development*. We sincerely thank Dr Koenraad Veys for chairing the scientific committee, and all the committee members who gave so generously of their time and expertise in putting together an excellent meeting.

The meeting once again attracted researchers, clinicians and patient representatives from multiple countries and generated overwhelmingly positive feedback.

The workshop provides opportunities for researchers to share findings, build collaborations and develop new ideas. Importantly, it also facilitated meaningful interaction between scientists and the cystinosis community, helping to ensure that research priorities remain aligned with patient needs.

Several new research collaborations were initiated following discussions at the workshop, demonstrating its continued importance as a catalyst for research progress.

The workshop celebrated the outstanding contributions of early-stage career researchers with the awarding of two prizes. **The Prof Roz Anderson Memorial Prize for Best Scientific Poster** was

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awarded to Wiktor Palka, an undergraduate student working with Dr Jennifer Hollywood in UCC presenting her work “Does the Ctns KO Rat Exhibit Muscle Dysfunction?” Wiktor is the first undergraduate winner of this prize. The **Cystinosis Ireland Prize for Best Oral Presentation to a Non-Scientific Audience** was awarded to Dr Sante Princiero Berlingiero from Emma Children’s Hospital, Amsterdam who introduced his work on “Improving energy metabolism in cystinosis kidney cells”. Sante was the winner of the 2024 Prof Roz Anderson Memorial Prize, and we are delighted to see his ongoing commitment to cystinosis research.

Planning also began for DCW 2026, which will take place as a streamlined online event due to the CNE International Conference being hosted in Dublin later in the year.

Awareness

Awareness activities continued to grow throughout 2025 under the leadership of Rachael Reilly and the Awareness Committee.

A key development was the expansion of our volunteer communications team, with Lucy Beckwith and Sinéad Maguire taking on an increasingly active role in content creation and social media engagement.

The committee developed and implemented:

- Social Media Guidelines
- Communications Policy
- Complaints Policy

These policies help ensure that our communications remain accurate, consistent and aligned with organisational values.

Throughout the year, our social media channels featured a mixture of personal stories, awareness campaigns, event updates and research content. Increased engagement demonstrated the value of sharing authentic experiences from individuals and families affected by cystinosis.

The committee also began developing more targeted communications for professional audiences through LinkedIn and other platforms, highlighting research developments, funded projects and scientific events.

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Work commenced on longer-term communications planning, including content calendars, campaign-based approaches and improved measurement of engagement and impact.

Strategy and Governance

As a regulated charity, we continue to place a strong emphasis on governance, transparency and accountability.

Throughout 2025 the Board invested considerable time reviewing and strengthening our governance structures.

Significant achievements included:

- Completion of an extensive review of our strategic plan.
- Development of a new Strategic Plan for 2026-2029.
- Ongoing review and enhancement of organisational policies.
- Completion of a Board Competency Review.
- Participation in a BoardMatch event to support future board recruitment.
- Development and maintenance of a comprehensive organisational risk register.
- Establishment of a Finance and Risk Committee.

The organisation also completed its annual return to the Charities Regulator and continued its commitment to operating in line with the Charities Governance Code.

A cyber-security breach was identified and managed during the year. The incident was reported appropriately, investigated by external specialists and resulted in additional security measures including enhanced multi-factor authentication. No sensitive information was compromised.

Externally, Cystinosis Ireland continued to engage with national and international organisations including Rare Diseases Ireland, Health Research Charities Ireland, EURORDIS, the Irish Kidney Association, Cystinosis Network Europe and a wide range of patient advocacy networks.

Finance

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Cystinosis Ireland continues to rely on the generosity and commitment of our families, supporters, volunteers, donors and corporate partners.

Our fundraising target remains €100,000 annually. Through a combination of community fundraising, corporate donations and individual giving, we once again progressed strongly toward this target in 2025.

Fundraising activities included:

- Grange Golf Day
- Ashbourne Golf Day
- Leixlip Tennis Event
- Mia's Bracelet Fundraiser
- Family-organised events in Cork and elsewhere
- Corporate donations and matched giving initiatives

Additional support was received from long-standing corporate and philanthropic partners who continue to make it possible for Cystinosis Ireland to invest in research and family support while maintaining strong operational governance.

The Board remains committed to prudent financial management, maintaining reserves, and ensuring all research funding commitments can be met sustainably.

Staffing

Our staff team comprises:

- **Denise Dunne, Operations Manager**
- **Dr Tracey McCauley, Research Manager**

Throughout 2025 Denise and Tracey continued to deliver the organisation's strategy, support families, manage research funding programmes, strengthen governance systems and represent Cystinosis Ireland nationally and internationally.

Their work has played a significant role in maintaining the organisation's reputation as a respected leader within the global cystinosis community.

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Future Planning

As we look toward 2026 and beyond, several important priorities have emerged.

The hosting of the 5th Cystinosis Network Europe International Conference in Dublin represents a major opportunity to strengthen international collaboration and support the global cystinosis community.

Other areas of focus include:

- Participation in the HRCI-HRB Joint Funding Scheme
- Expansion of Seedcorn research funding
- Engaging with and improving supports for adults living with cystinosis
- Continued development of support for siblings of people with cystinosis
- Exploration of newborn screening opportunities
- Adult healthcare and transition services
- Enhanced measurement and reporting of organisational impact

As always, our future work will be guided by the needs and priorities of people living with cystinosis and their families.

We welcome your ideas, feedback and involvement as we continue working towards better treatments, improved support and ultimately a cure for cystinosis.

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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 2025, 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).

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

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 2025

In 2025, 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

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online. At our December 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. One adult with cystinosis attended.

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
Jan	✓	x	✓	✓	x	x	✓	x	✓
Mar	✓	x	✓	✓	✓	✓	x	✓	✓
May	✓	✓	✓	✓	x	x	✓	✓	✓
Jun	✓	✓	✓	✓	x	✓	x	x	✓
Sep	✓	✓	✓	✓	✓	✓	x	x	✓
Oct	✓	✓	✓	✓	✓	x	✓	✓	✓
Dec	✓	✓	x	✓	✓	✓	✓	✓	✓

Research - Awareness - Support

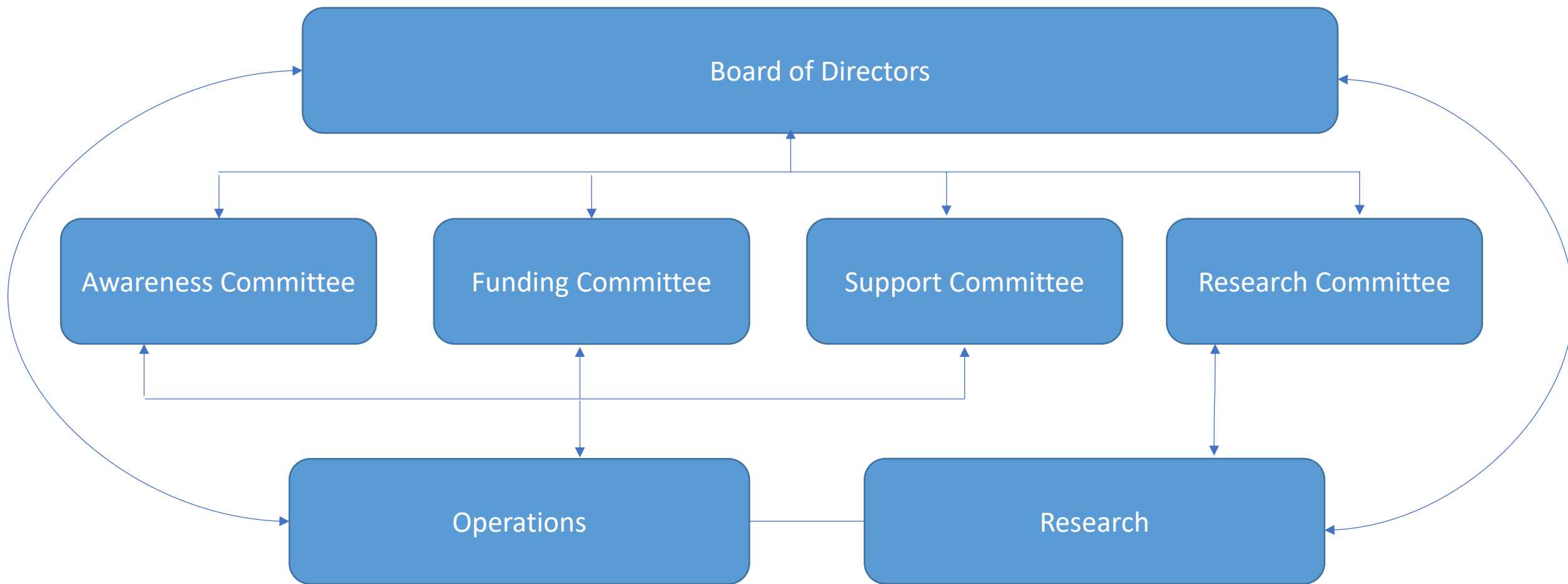
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cystinosis
ireland

Awareness

Rachael Reilly (Chair)
Liam McFadden
Karen McCullagh
Lucy Beckwith
Sinéad Maguire

Support

James Ennis (Chair)
Sue Maguire

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

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