



## CHAIR'S REPORT

2023 was an important year for Cystinosis Ireland. We welcomed the return to a more normal world with open arms and we marked the 20<sup>th</sup> anniversary of our organisation. Twenty years ago, a cystinosis diagnosis was a very different prospect to one received today. Cysteamine therapy was available and research being conducted but over the last two decades we have seen a significant shift in the potential therapies being identified through research. In Ireland, people living with cystinosis now have two forms of therapy available as well as access to many more supports for living better with cystinosis. The research community continues to work on many other options, some of which may offer improved quality of life as well as mitigating against the impacts of the disease.

Cystinosis Ireland is very proud of the work we have done in those 20 years investing in the best research internationally, supporting families through our formal and informal networks, representing the Irish community in international projects and supporting the formalisation of Cystinosis Network Europe which brings the international community together. This summer we marked our investment since inception of €3m in research, either as a sole or co-funding partner. This was a significant cause for celebration when we met with our families in Barretstown. This fundraising has come from our supporters – all of the core costs of the organisation are supported by a philanthropic donor, so all donations are used to fund research and to support people living with cystinosis, their families and carers.

As we look towards the next 10 years and the next generation of Cystinosis Ireland, we invite all of our families and supporters to become involved in our work. If you have ideas or skills you can share, please do. We welcome your engagement at whatever time and pace suit you. Cystinosis Ireland is volunteer led and we do the work we do with the support and involvement of those who matter most – people living with cystinosis.

### Research

Throughout 2023 Cystinosis Ireland continued to build our research portfolio.

In 2022 Cystinosis Ireland was successful in securing co-funding for two projects through the HRCI-HRB Joint Funding Scheme. These projects led by Dr Jennifer Hollywood in Auckland, New Zealand, and Prof Elena Levchenko, KU Leuven, Belgium began in 2023.

A stand-alone project by Prof Joyce Senior in UCD began also. This project will support children and young adults in understanding their educational support needs and help provide guidance in accessing supports through the educational system. This psychosocial area of research is new to our research portfolio and an area where we are excited about being able to bring tangible benefits to people in our community. The research team joined the family meeting in Barretstown in April to meet our families and begin their work. This was a really important opportunity for both the research team and the families to understand the potential impact of this type of research.

### *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](http://www.cystinosis.ie)  
[facebook.com/cystinosisirl](https://facebook.com/cystinosisirl)  
[twitter.com/CystinosisIrl](https://twitter.com/CystinosisIrl)  
[Instagram.com/cystinosisireland](https://Instagram.com/cystinosisireland)  
[mail@cystinosis.ie](mailto:mail@cystinosis.ie)

In Ireland, the Human Tissue Bill continued to work its way through the legislative process this year. We anticipate it being signed into legislation in 2024. This legislation is vital as it will pave the way for soft-opt out organ donation. Cystinosis Ireland is a member of the Irish Donor Network, a network of patient advocate organisations, which has campaigned for many years for this legislative change. Soft opt-out is the standard in many European countries and has the potential to greatly increase the numbers of donor organs available. We look forward to the bill's enactment.

### Research and family event

Cystinosis Ireland held the 9<sup>th</sup> annual Dublin Cystinosis Workshop and Family Workshop on 13 – 16 April 2023 in Barretstown Camp, Co Kildare. The events also marked our 20<sup>th</sup> anniversary. The weekend was a huge success with more than 100 members of our community – patients, families, researchers and scientists – coming together to share experience, learn and have some fun.

The Dublin Cystinosis Workshop is an interactive forum for researchers to share knowledge, spark new ideas and explore new research approaches aimed at better understanding and treating cystinosis.

2023's themes were:

- 1) Evolution in mechanistic / pathogenic insights in cystinosis,
- 2) Innovative treatment approaches/ Innovation in treatment and
- 3) Novel clinical aspects.

The Workshop heard from researchers and clinicians from the USA, Canada, New Zealand, UK and Europe (including Ireland) who shared 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. As part of our poster presentation, Louise Medaer of Katholieke Universiteit Leuven, was awarded the Professor Roz Anderson Memorial Prize for her presentation of her work on S Isogenic human cell models to unravel the underlying mechanism of cystinosis myopathy.

The Family Workshop was fun filled and action packed. Adults living with cystinosis, parents and caregivers were given the opportunity to engage with our research and clinical colleagues to learn more about areas of importance to the community including therapy development, new areas of research and potential treatments of the future. Our families also engaged with our research community to discuss current research projects and gain a greater understanding of the research process. Our up-and-coming researchers were challenged to explain their work to a lay audience, who voted on the best presentation.

The Cystinosis Ireland Award for Best Oral Presentation to a Non-Scientific Audience was awarded to Hayley Chang of the Frederick J. and Marion A Schindler Cognitive Neurophysiology Lab (CNL), University of Rochester School of Medicine and Dentistry, New York, for her work on Development and Characterization of a Cystinosis Knock-Out Mouse Model Using CRISPR/Cas9.

### 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](http://www.cystinosis.ie)  
[facebook.com/cystinosisirl](https://facebook.com/cystinosisirl)  
[twitter.com/CystinosisIrl](https://twitter.com/CystinosisIrl)  
[Instagram.com/cystinosisireland](https://Instagram.com/cystinosisireland)  
[mail@cystinosis.ie](mailto:mail@cystinosis.ie)

While the adults were learning and sharing their perspectives with the scientific attendees, the children were enjoying activities including archery, fishing, canoeing, wall climbing, crazy golf, movie making and prank parties. The sun shone and screens were abandoned for an opportunity to make new friends and try new experiences. The Barretstown team were invaluable in supporting the children to enjoy every moment of the weekend.

Cystinosis Ireland is very grateful to all involved in putting this weekend together: the DCW Scientific Committee, under the guidance of Dr Koenraad Veys; our Support Committee; and our staff, as well as the staff and volunteers at Barretstown.

## Support

Adults living with cystinosis and families of everyone living with cystinosis continue to be the central focus of our work in Cystinosis Ireland.

We were deeply saddened at losses of members of our community in 2023. Darren Barnes passed away in January and William (Billy) McCauley in August. Both are sadly missed by their loving families, and we offer our sincere condolences to all. May they rest in peace.

In 2023 a family including two adults with cystinosis moved from the UK to Ireland and joined our Irish community. The peer support provided by our closed Facebook group is a key resource, as is the personal connection between members of our board and staff with our families. Cystinosis Ireland offers a support network across the island of Ireland, and we work with our colleagues internationally to support people with cystinosis and their families where we can.

In 2023 Cystinosis Ireland offered a new mental health resource to our community. Working with an online counselling service, we offer an opportunity to avail of up to 12 sessions with an accredited counsellor. Details of how to access this service are available from our support committee - [support@cystinosis.ie](mailto:support@cystinosis.ie).

Our support committee provided financial aid to families with cystinosis to attend our research and family meeting in Barretstown to allow as many as possible to engage with our research and clinical community as well as to have a fun time in a safe and engaging environment. It was also an important opportunity for the children, teens and adults with cystinosis to get to know each other and build their own community.

## Awareness

During 2023, while remembering the work of our last 20 years, the awareness committee worked hard to develop a plan for communicating into the future, a key component of which will be a new website to be launched in 2024.

## *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

Our social and other media continued to provide outreach, education and support to our community both in Ireland and internationally, sharing our stories and milestones with humour and grace.

From Rare Diseases Day in February to fundraising and awareness events, our social and other media is a valuable tool to connect and support us all.

### **Fundraising**

Fundraising is key to our sustainability to invest in research and supporting people living with cystinosis.

We were delighted that our long-standing golf events at the Grange and Ashbourne Golf Clubs were back to their traditional formats. We are as ever humbled by the generosity of our supporters in their time, money and expertise in supporting our work through our fundraising events. Similarly, our friends in Leixlip Tennis Club gave their time and dedication for their annual event, as did the Carrigaline Ladies' Day event which was a huge success.

Families and friends continued to arrange Facebook fundraisers for birthday and memorial occasions. We are constantly surprised by the innovative ways in which our supporters take on challenges and innovative fundraising ideas.

### **International**

Cystinosis Ireland continues to be a leading organisation internationally, supporting research and our community.

We continue to provide the secretariat to Cystinosis Network Europe (CNE) and are represented on that organisation's board of directors, including Anne Marie O'Dowd, being Chair both of CNE and its Cystinosis Community Advisory Board (CAB). Our director Niamh O'Brien also served as a board member and Denise Dunne acts as Secretary of CNE.

The Cystinosis CAB is a representative group of trained patient advocates who engage with the research community (clinical, lab based and commercial) in an effort to identify research priorities and support the best possible research outcomes for our community. This international engagement ensures Cystinosis Ireland is at the forefront of research, support and information as it emerges and gives us access to a world class public patient involvement (PPI) panel to ensure the research we support is as meaningful as possible for our community.

### **Strategy and Governance**

Cystinosis Ireland has agreed a three year strategy for our organisation which will be reviewed and updated on a rolling basis.

As a regulated charity we understand the importance of being transparent in everything we do and we subscribe to continuing to be a best in class organisation.

### *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.*

We completed our annual return to the Charity Regulator in October 2023.

Our board works on an ongoing basis to ensure the high level of all the work we do is reflected in our processes.

In 2023 we revised our audit and accounting processes to adhere to the Financial Reporting Standard 102 (FRS 102). While our previous accounting practice was completely transparent and met all required standards, the FRS 102 is recommended by the Good Governance Code, and we decided to implement it in our reporting.

### 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.

### In Conclusion

Reflecting on the 20 years of our organisation has been a humbling and invigorating opportunity. People living with cystinosis have seen great strides in therapy and research in that time and we are proud to have been part of that work through our investment in research and in supporting the development of resources for those living with the condition. It is important to acknowledge the incredible amount of work our voluntary board has provided in that time. They give their time and expertise generously and completely and with endless passion to improve the lives of those living with cystinosis.

Our family and scientific event in Barretstown was a true celebration of our work and we were delighted to be able to report the investment to date of €3m in research projects focused on all aspects of this disease, either through direct funding or as a co-funding partner. For an organisation of our size in an ultra rare disease area, we are immensely proud of this significant contribution to the research community.

The work of Cystinosis Ireland is focussed on Research, Education/Awareness and Support and we hope we live up to these in our work we do every day.

Thank you to everyone who is such an important part of our work.

### *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](http://www.cystinosis.ie)  
[facebook.com/cystinosisirl](https://facebook.com/cystinosisirl)  
[twitter.com/CystinosisIrl](https://twitter.com/CystinosisIrl)  
[Instagram.com/cystinosisireland](https://Instagram.com/cystinosisireland)  
[mail@cystinosis.ie](mailto:mail@cystinosis.ie)

Mick Swift  
Chair  
19 September 2024

### *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](http://www.cystinosis.ie)  
[facebook.com/cystinosisirl](https://facebook.com/cystinosisirl)  
[twitter.com/CystinosisIrl](https://twitter.com/CystinosisIrl)  
[Instagram.com/cystinosisireland](https://Instagram.com/cystinosisireland)  
[mail@cystinosis.ie](mailto:mail@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.

### Our vision

We strive for a cure for cystinosis. Until then, we want people living 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 2023, those who served on our board were: James Ennis; Andy Maguire; Sue Maguire; Karen McCullagh; Thomas McDonald; Liam McFadden; Niamh O'Brien; Anne Maire O'Dowd; Rachael Reilly; and Mick Swift (Chair).

The Cystinosis Ireland board is comprised of nine individuals with an interest in cystinosis personally or professionally. Niamh O'Brien resigned from the board in September 2023 and we thank her for her insight and engagement during her tenure.

The board is supported by our operations and research managers. As set out in our strategic plan we have four working groups – Awareness, Funding, Research and Support. Each of the groups is chaired by a board member. The

### *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

working groups provide an annual workplan and report on their activities at each board meeting. The working groups 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. A visual representation of our organisational network is below.

A key consideration in the work of our governance review was 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 patient 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 cystinosis. 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. We welcome members of our community and those with industry expertise to offer to join the team in whatever capacity is possible.

## Board meeting attendance 2023

In 2023, the board met on 5 occasions. Three 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 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	L McFadden	AM O'Dowd	N O'Brien	R Reilly	A Maguire	S Maguire	M Swift
February	✓	x	✓	✓	✓	✓	x	x	✓
March	✓	✓	✓	✓	x	x	✓	✓	✓
June	✓	✓	✓	✓	x	✓	x	✓	✓
September	✓	✓	✓	✓	resigned	✓	✓	✓	✓

## 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.





November	✓	✓	✓	✓	-	✓	✓	✓	✓
----------	---	---	---	---	---	---	---	---	---

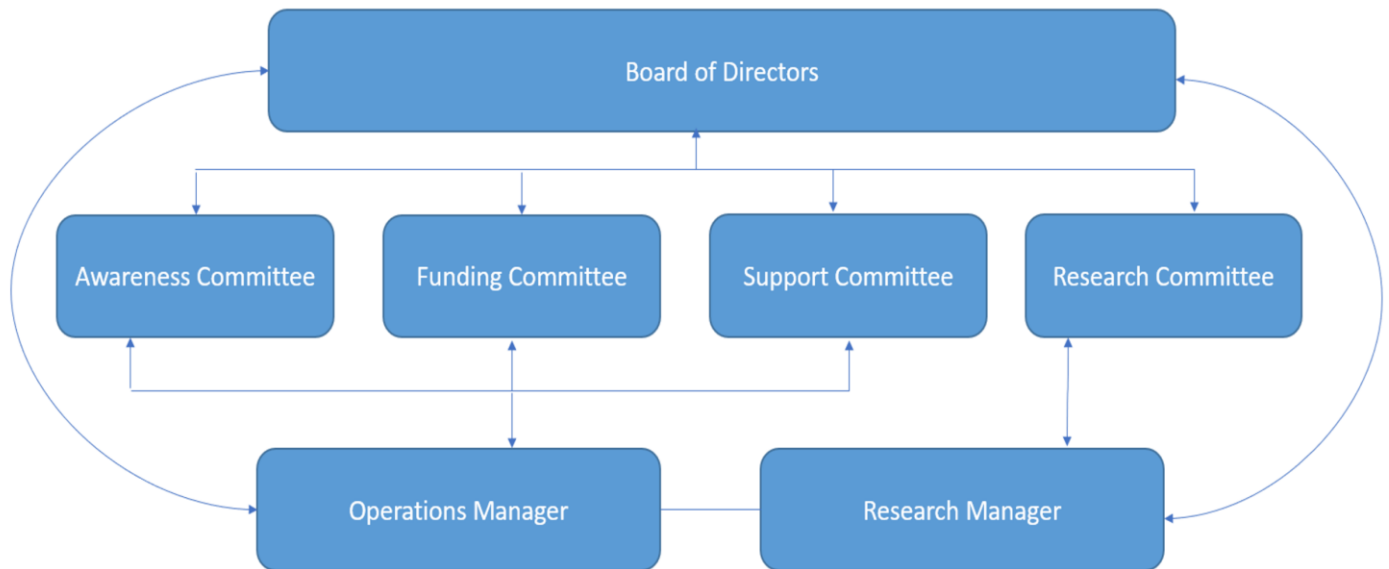
### Staff and Volunteers

In 2023, we had two staff members, Denise Dunne looking after our operations and Dr Tracey McCauley managing our research work.

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.

### 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.



## *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.