



cystinosis
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CHAIR'S REPORT

As the world began to open up again in 2022, we breathed a collective sigh of relief that our work could once again look further out and into our community.

Research

Throughout 2022 Cystinosis Ireland continued to build out our research portfolio.

The HRB/HRCI Joint Funding Scheme opened and Cystinosis Ireland was delighted to be funded for both applications we submitted. These projects will be led by Dr Jennifer Hollywood in Auckland, New Zealand, and Prof Elena Levtschenko, KU Leuven, Belgium. The scheme was highly competitive as always and Dr Hollywood's was the top ranked projects of the 15 ultimately funded. We sincerely thank our Research Committee and peer reviewers for their thorough and committed work to ensure the investment we make in research is in the best and most effective.

In addition to these projects, Cystinosis Ireland committed to funding a research project led by Prof Joyce Senior in UCD which 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 annual Dublin Cystinosis Workshop was held online on Thursday 21st April 2022, creating collaborations and conversations between world class researchers and clinicians. Now in its eighth year, this unique event allows those active or interested in cystinosis research to share 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. Sixty six attendees heard 20 presentations with key areas of discussion including the odour created by therapies for cystinosis; fertility; and potential novel kidney transplant protocols. The meeting was also addressed by an expert in Public Patient Involvement (PPI), an area in which we endeavour to always meet best research practices.

The Prof Roz Anderson Memorial Prize for the best short science communication was awarded to Elena Sendino Garv. Elena is a PhD candidate at the Utrecht Institute for Pharmaceutical Sciences (UIPS), and is working on kidney ciliopathies and tubulopathies, with the aim of using drug repurposing approaches and gene-editing (Crispr-Cas9) to restore the cell function of these kidney conditions.

The Workshop also took the opportunity to celebrate the tremendous work by Dr Don Cairns and Dr Bill Gahl and the contributions they have made to cystinosis research and patient communities in their careers. We also took the opportunity to pay tribute to the late Dr Jerry Schneider who is sadly missed.

Research - Awareness - Support

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

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We offer our sincere thanks to the members of the organising committee for the DCW 2022, ably led by Anne Marie O'Dowd who chairs our Research Committee and also to Dr Tracey McCauley who joined us this year as Research Manager to further add to our capacity.

Support

Our families continue to be the central focus of our work in Cystinosis Ireland.

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 and families. Our support network extends the length and breadth of the island of Ireland, and we work with our colleagues internationally to support people with cystinosis and their families where we can.

In late November, the Irish cabinet approved the Human Tissue Bill which 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 its continued passage into legislation.

In 2022 our support committee provided financial aid to families with cystinosis to travel to the CNE International Conference in Leuven and other significant events. These grants are an important resource to facilitate access to learning about cutting edge research and meet with members of our international scientific and clinical world as well as families from all over the world who have similar life experiences of cystinosis. We continue to encourage our families to avail of this support.

2022 also saw the beginning of an adults with cystinosis peer support network building across Ireland. This is co-ordinated by James Ennis who leads our Support Committee.

Awareness

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 when we cannot be together in person. Our Awareness Committee is led by Rachael Power.

Fundraising

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

We were delighted that our traditional golf events in Grange Golf Club and Ashbourne Golf Club were able to resume in more normal conditions. Our after course events were curtailed, but we enjoyed the camaraderie of being on the

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beautiful courses, raising funds and awareness for our cause. 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 over their time and dedication for their annual event.

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. We offer our huge thanks to Aaron Byrne who was one of a number of people to shave his head in Ashbourne Golf Club in October. Board members Andy and Sue Maguire are invaluable in ensuring our fundraising events are such successes.

International

As the world reopened, the CNE International conference was held in July by our colleagues in the Dutch-Flemish Cystinosis Group.

The conference was in Leuven, Belgium, and was chaired by Prof Dr Elena Levchenko, an expert in the field both as a clinician and a researcher. The Irish contingent was strong with members of our board and staff attending along with some of our family members. The conference was an important touch stone after Covid, allowing our wider, mainly European, community to gather and make valuable in person connections. The conference highlighted work that needs to be done to support our international families in accessing therapies that are standard in many European countries.

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.

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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.

We completed our annual return to the Charity Regulator in October 2022

Our board worked extensively following on our strategy and governance through the Covid restrictions, we continue this work to ensure the high level of all the work we do is reflected in our processes.

Staffing

Our staff at cystinosis Ireland is comprised of Denise Dunne, Operations Manager and Dr Tracey McCawley, Research Manager.

Dr Tracey McCawley joined the team in July 2022. Tracey has a background in exercise physiology and extensive experience in managing a research portfolio. As our investment in cutting edge research in cystinosis, her expertise will be a valuable resource.

Tracey took on the role vacated by Dr Nora Lieggi who left us this year. Nora contributed significantly to our research work through her extensive experience in public and patient involvement (PPI), a core value of our research ethos.

In Conclusion

Since our establishment in 2003, Cystinosis Ireland has invested in research projects focused on all aspects of this disease to the value of in excess of €2 million 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.

We looking forward to marking our 20th anniversary in 2023 and celebrating the many achievements of the community during that time.

Thank you to everyone who is such an important part of our work. Our Board is entirely voluntary and we could not achieve a fraction of what we do without their knowledge, expertise, drive and endless passion to improve the lives of those living with cystinosis.

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

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Mick Swift
Chair
14 September 2023

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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 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 2021 began the work of solidifying our compliance with the Charities Governance Code.

The work begun in 2020 on our strategy and the board continues to implement the necessary changes and strengthen the areas where we identified weaknesses.

While the responsibility for governance is a collective one, the work itself was driven by Denise Dunne and Mick Swift from an operational perspective.

Our Board

The Cystinosis Ireland board is comprised of nine individuals with an interest in cystinosis personally or professionally. Our board members are: James Ennis; Andy Maguire; Sue Maguire; Thomas McDonald; Liam McFadden; Niamh O'Brien; Anne Maire O'Dowd; Rachael Reilly; and Mick Swift (Chair).

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The board is supported by our operations and research managers.

Following a strategic and governance review process in 2020/2021 we now have four working groups – Awareness, Funding, Research and Support.

Each of the groups is chaired by a board member. The 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 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 expertise to offer to join the team in whatever capacity is possible.

As part of our work in 2022, the Board developed a handbook to address the role and responsibilities of those appointed and in 2023 will begin a process of evaluating where current skills are focussed and where we may require additional expertise.

Board meeting attendance 2022

In 2022, the board met on 7 occasions. Five meetings were held online, with the September and November meetings being hybrid – those who could not attend in person were invited to join online.

The attendance at meetings is set out below.

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

	J Ennis	T McDonald	L McFadden	AM O'Dowd	N O'Brien	R Reilly	A Maguire	S Maguire	M Swift
February	√	√	√	√	√	√	x	x	√
March	√	√	√	√	√	√	x	x	√
May	√	√	√	√	x	x	√	x	√
June	√	√	√	√	x	√	√	√	√
September	√	√	√	√	√	√	√	√	√
October	√	√	√	√	x	x	x	√	√
November	√	√	√	√	√	√	x	√	√

Staff and Volunteers

In 2022, we had two staff members, Denise Dunne looking after our operations and for the first half of the year, Nora Lieggi managing our research work. Nora finished working with us in July and was replaced by Tracey McCauley.

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.

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