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## Cystinosis Ireland Annual Report 2020

2020 was an unusual and difficult year for many in our community.

As we attended the event to celebrate International Rare Disease Day in Belfast, the first case of Corona Virus, Covid 19 was reported in Ireland. The unexpected lockdown hit the country quickly and the landscape changed immeasurably. It also brought with it new ways of engaging with our international peers and families and in many ways brought us closer.

In a year of change and uncertainty, we are proud of what we achieved.

Through 2020, our Board and our team worked to deliver on our goals and commitments through a unique and challenging environment. The Board is supported by our staff comprised of Dr Ruth Davis leading our research and Denise Dunne looking after our operations.

Despite the potential for the global pandemic to halt activities, instead we faced the challenges head on, with our work expanding and becoming more complex as the months passed. Our reputation and impact in cystinosis research and the wider community continues to grow and is enhanced by our constant commitment to excellence.

### 2020 CNE International Conference

In 2018, Cystinosis Ireland was selected by Cystinosis Network Europe (CNE) to host the 2020 CNE International Conference to take place in Dublin in July 2020. When the world began to lock down in early March, we took the significant and ultimately very successful decision to host the meeting as a virtual event using the Zoom online platform on 25<sup>th</sup> April.

The success of the day took everyone by surprise.

In the space of six weeks, the organising committee transformed our planned in person event to an online day reaching more than 600 participants in 49 countries, across 20 time zones and in eight languages. As the largest cystinosis event ever held, we were amazed with the connectedness we achieved and it has been an important lesson for us to remember those who, for many reasons, cannot travel to join events in person.

The Virtual Conference marked the start of what we hope will be a more extensive online outreach to families everywhere. The presentations from the conference, in English and translations, are available on the Cystinosis Ireland website.

### *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, T. McDonald, L. McFadden, N. O'Brien, AM. O'Dowd, R. Reilly, M. Swift.

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Following the success of the Virtual Conference, we decided to build on our online outreach and held the 6<sup>th</sup> Dublin Cystinosis Workshop online in July.

This meeting again allowed a wider reach to clinicians and scientists who may not have otherwise been able to travel to Dublin. It allowed us to gain new insights into the difficulties facing our research community during the global pandemic and the work they were continuing to produce during this time.

Although the event could not provide the social and engagement opportunities of previous years, we were delighted to be joined by researchers across the world, from New Zealand to California. Dr Jennifer Hollywood won this year's Professor Roz Anderson Memorial Prize which was awarded for the best short communication presented at the workshop. Dr Hollywood was awarded the prize following a direct poll of her scientific peers participating in the workshop.

## Research

Internationally, research is developing significantly in spite of the Covid-19 pandemic closing some labs and curtailing some research activity.

Dr Stephanie Cherqui and her team continued to work on their gene therapy clinical trial in California. This potentially life-changing therapy offers cautious hope to many individuals living with cystinosis and we look forward to seeing how this study progresses.

Prof Herbie Newell continues the work begun by Prof Roz Anderson on a pro-drug which is a cause for optimism.

Prof Katharina Hohenfellner started a more limited than hoped, but important study into the importance of exercise for people with cystinosis. While these pieces of research stand out, there is a significant body of work being done in the field which will provide if not a cure, then potentially improved therapies to increase the daily quality of life for those living with cystinosis.

## Cystinosis Network Europe (CNE)

Cystinosis Ireland continues to be a leading organisation internationally supporting research and our community. CNE was formalised in Dublin in 2020 with Anne Marie O'Dowd taking on the role of Chair both of CNE and its Cystinosis Community Advisory Board (CAB). The Cystinosis CAB is a representative group of trained patient advocates who engage with the research community (clinical, lab base and commercial) in an effort to identify research priorities and support the best possible research outcomes for our community.

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Cystinosis Ireland hosts and provides the secretariat for CNE. This international engagement ensures Cystinosis Ireland is at the forefront of research, support and information as it emerges.

## Strategy and Governance

In order to ensure the work we do continues to be relevant and meet the needs of our community, we took the opportunity in late 2020 to conduct a strategic and governance review.

With the support of Sandra Velthuis and Sheila Cahill we had input from our families, researchers, peer organisations, and the wider research community to identify where we are working well and where we need to refocus our efforts. As part of the governance review we identified that those sitting on our Executive Committee should be formally appointed as company directors. We thank all of our committee for agreeing to formally take on this responsibility - James Ennis (NI), Andy Maguire, Sue Maguire, Tom McDonald, Liam McFadden, Niamh O'Brien, Anne Marie O'Dowd, Rachael Reilly, and Mick Swift.

We thank all who gave their thoughts and shared their views during this process on how we can support research and our families better. While the strategic review has been completed for this cycle, we continue to welcome any feedback on how we do our work. The guiding ideals of Cystinosis Ireland are Research, Education and Support and we hope we live up to these in the work we do every day.

## Investing in Research

We were delighted to mark investing €2million in research during 2020.

Since its establishment in 2003, Cystinosis Ireland has supported research projects focused on all aspects of this disease to the value of €2 million either through direct funding or as a co-funding partner. Since its inception in 2006, Cystinosis Ireland has engaged with the HRB-HRCI (then MRCG) Joint Funding Scheme. Through it, we have been able to maximise the amount of funding we could leverage to invest in research, with our 50% contribution being matched by the HRB fund following a rigorous peer review process. Cystinosis Ireland is proud to be one of the most funded charities in the HRCI group to have projects funded through this scheme. We have used a similar model in working with our colleagues in Cystinosis Research Network (US) and Cystinosis Foundation UK to co-fund projects more recently.

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 couldn't do it, however, without the incredible efforts of our supporters who fundraise for us.

RESEARCH - AWARENESS - SUPPORT

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2020 was a very difficult year for fundraising with events cancelled and social distancing making gatherings of any type nearly impossible. We offer huge thanks to those who continued to take on small, personal challenges. The dedication of our families to each other and the work we do is awe inspiring.

### Graham Barnes

The end of the year was a sad one, with the passing of Graham Barnes. Graham was one of the first people in Ireland to be diagnosed with cystinosis, before the treatments we have now were available. Graham was remembered by his brother Clive at his funeral as a funny and kind man, a keen football supporter and a musician who was greatly loved. We continue to offer our sincere condolences to all who loved Graham, in particular Catherine, Michael, Clive and Darren. Rest in peace.

We were also contacted by a family of a newly diagnosed child and, as an organisation and a community, we offer our support to those who need and want it.

We are aware of the hard work that has gone into 2020 for everyone, and we thank you for your continued support.

Mick Swift

Chair

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## SUPPORT AND EDUCATION

With the 2020 lockdowns, in person support was a challenge. Sue Maguire continued to be our primary point of contact for our families by telephone, and our family Facebook page was valuable for parents to support each other.

James Ennis continued to work with our families in Northern Ireland and bring their perspective to our discussions at Board level.

Because of the pandemic, most education and support events moved online removing the important face to face contact with our international friends and colleagues but through CNE's Virtual International Conference, we were able to keep in touch with emerging research developments and extend an online connection to families worldwide.

## FUNDRAISING

Cystinosis Ireland has an incredible number of supporters who take on fundraising challenges for us every year. These efforts were severely curtailed in 2020. We were delighted that those who were in a position to take on individual challenges still did, and our corporate supporters maintained their valued commitment to our work.

## ADVOCACY

Cystinosis Ireland is registered on the Register of Lobbyists where our advocacy work can be seen (<https://www.lobbying.ie/>).

We continue to support the progress of the Human Tissue Bill and "soft opt-out" for organ donation, an issue close to the hearts of our members.

To support our work, Cystinosis Ireland is represented on and engages with a number of national and international organisations –

- Health Research Charities Ireland (formerly Medical Research Charities Group), is an umbrella organisation for medical research and patient support charities. It is through the HRCI that Cystinosis Ireland has been able to access to the Joint Funding Scheme which has been hugely significant in leveraging our research funding work for many years.
- Irish Platform for Patient Organisations, Science and Industry (IPPOSI) is a patient-led organisation that works with patients, government, industry, science and academia to put patients at the heart of health policy and innovation.
- Rare Disease Taskforce brings together groups working in this area and has been heavily involved in the Rare Disease National Plan development and implementation.
- Rare Disease Technology Review Committee incorporates the patient and clinical perspective in the process of medicine reimbursement and approval.
- Organ Donor Network is a group of organisations working in the area of organ donor and transplant. The work of the last year has been mainly towards lobbying to implement the 'soft opt-out' system.
- Rare Disease Ireland is a patient advocacy national alliance for voluntary groups representing people affected by or at risk of developing a rare disease.
- EURORDIS, the European Rare Disorders Organisation
- Cystinosis Network Europe

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## INTERNATIONAL ENGAGEMENT

In 2020 Cystinosis Ireland officially took on the Chair and leadership of CNE to promote the interests of the cystinosis community internationally. Membership of CNE is broad and includes representation from cystinosis patient organisations in Europe, Australia, South Africa and the US.

Cystinosis Ireland provides the secretariat for CNE with Anne Marie O’Dowd serving as the Network’s Chair. Under her stewardship the group has continued to work on developing the Cystinosis CAB. This patient advocate led group will be the main contact point for our network to discuss research, patient engagement, and potential trials and therapies with pharmaceutical companies and early stage researchers. The structure of the Cystinosis CAB, will facilitate discussions (in a neutral setting) on the latest developments and challenges related to medical research and procedures with the companies or bodies conducting the research.

The Cystinosis CAB continues to engage with a wide range of researchers and companies to promote the need to value the patient voice and value patient expertise at all stages of the research process.

## CYSTINOSIS IRELAND RESEARCH 2020

This has been another exciting year for Cystinosis Ireland research, yet it was also one that brought significant challenges linked to the impact of the Covid 19 pandemic both on our own activities as well as those of our researchers worldwide.

### New research funding awarded by Cystinosis Ireland in 2020

Although the temporary closure of laboratories as a result of various lockdowns delayed progress on some of our ongoing research projects throughout 2020, we still managed to grow our research programme during this turbulent time.

In 2020, the Cystinosis Ireland Board approved funding for three new research projects:

- Literature review of recent cystinosis research by Michael Deane, University College Cork, IE – Award Amount: €4,356 (Summer studentship under the supervision of Dr P Harrison, UCC and Dr J Hollywood, U of Auckland, NZ)
- IMPACT Study – Dr Katharina Hohenfellner, Klinik für Kinder- und Jugendmedizin – Kinderneurologie, DE – Award Amount: €20,000
- Biomaterial-mediated delivery of CTNS gene for ocular cystinosis – Dr Valeria Graceffa, Institute of Technology, Sligo, IE – Award Amount: €9,990

In addition, we were thrilled to join forces with our USA based sister organisation, Cystinosis Research Network to co-fund on a 50:50 basis a research project hosted in Professor Minnie Sarwal’s laboratory in University of California, San Francisco. The project, entitled “*A Cellular Resource for Studying Male Infertility in Cystinosis*” and which has a total award value of €10,000, is led by post-doctoral research fellow, Dr Swastika Sur.

For more information on all of our funded research projects you can visit our website at: <https://cystinosis.ie/home/research/cystinosis-ireland-research-grants-awarded/>

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## KEY RESEARCH EVENTS IN 2020

### Virtual International Cystinosis Conference 2020

On behalf of CNE, Cystinosis Ireland hosted the first ever CNE Virtual International Cystinosis Conference 2020 on 25 April 2020. Within the space of just six weeks, CNE and Cystinosis Ireland took the disappointment of cancelling the highly anticipated International Cystinosis Conference 2020 (which had been scheduled to be held in Ireland in July 2020 in Dublin) and converted it into a hugely successful virtual event.

The virtual conference attracted an audience of more than 600 participants from 49 different countries spanning 20 time zones across the world including countries as far away as Australia, Mexico, Turkey, Egypt, USA and Canada. The diverse mix of attendees included adults and families with many years' experience of living with cystinosis as well as families that have newly diagnosed young children and are just beginning their cystinosis journey.

Many of the conference logins included several members of families. Many of those who joined would not have been able to travel to the original in-person conference that had been planned. One family in Australia listened in to the whole conference until 3 in the morning! Several participants noted that they had never before had an opportunity to listen to world class cystinosis experts speak and answer questions about their ultra-rare condition.

In order to address the needs of this diverse audience, the conference drew on a wide variety of expert contributors who focused on providing essential information about cystinosis as well as highlighting new knowledge and breakthroughs in our understanding and treatment of the disease. The conference was simultaneously translated from English into seven different languages and featured presentations on a wide variety of clinical and research aspects of cystinosis from 16 world renowned clinicians, researchers and other healthcare professionals from USA, Canada, Ireland, Belgium, Germany, France, England and Scotland.

The keynote speaker was Dr Stephanie Cherqui, Associate Professor at the Department of Paediatrics, Division of Genetics of the University of California, San Diego, USA. Dr Cherqui's highly anticipated contribution to the conference focused on her pioneering research developing an autologous stem cell and gene therapy for people living with cystinosis for which she received FDA approval for a human Phase 1/2 clinical trial in October 2019. Dr Cherqui provided an update on the positive progress of the first patient enrolled on this clinical trial who received modified version of their own stem cells which had been genetically engineered to produce and deliver functional cystinosin throughout the body. the future once circumstances permit.

One of the most important elements of the virtual conference for family participants, both new and experienced, was the hour-long Q&A session at the end during which questions submitted through the chat feature answered by the panel of cystinosis experts.

The positive feedback on the virtual conference from participants which came in live via the online chat function was both humbling and heart-warming. Participants were highly engaged, sought answers to many interesting questions and queried participation in various upcoming trials. One of our experts, all of whom are well used to speaking at major scientific conferences, noted that this was the largest audience to whom he had ever presented.

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Cystinosis Ireland and CNE are very grateful to all those who assisted in delivering this very important international conference including the members of the International Conference Scientific Committee, which was ably chaired by Professor Elena Levtchenko, to all of the speakers, clinical experts and researchers who contributed to the conference as well as to the sponsors of the event.

Cystinosis Ireland and CNE are incredibly proud of the success of this first ever virtual cystinosis conference and on setting a new standard for future such conferences in the cystinosis world.

You can find out more about the CNE Virtual International Cystinosis Conference 2020 and all of the contributors (including all of the presentations and translations) on the conference website at: <https://cystinosis.ie/international-work-2/virtual-international-conference-2020-resources/>

### Dublin Cystinosis Workshop 2020

Following the incredibly successful CNE Virtual International Cystinosis Conference which was held online in April 2020, Cystinosis Ireland, responded to calls from the cystinosis research community to host the 6th Annual Dublin Cystinosis Workshop 2020 virtually on 16 July 2020.

Unlike the International Cystinosis Conference which is family-focused, the Dublin Cystinosis Workshop is a scientifically focused meeting aimed specifically at the scientists, clinicians healthcare professionals and patient experts in the field of cystinosis treatment and research.

Despite Covid 19 bringing the world of in-person meetings to a halt, the Virtual 6th Annual Dublin Cystinosis Workshop 2020 welcomed our largest ever participant audience of 49 experts from 10 countries (Belgium, Netherlands, France, Italy, Germany, Ireland, UK, USA, Canada and New Zealand) to share recent advances in their respective research endeavours and to look at new ways to develop our understanding of cystinosis as a disease and its treatment.

In welcoming all of the participants to the workshop, Mr Mick Swift, Chair of Cystinosis Ireland reflected on the importance of maintaining links and the exchange of research information during Covid 19 pandemic. The virtual meeting was ably co-chaired by long time champions of Cystinosis Ireland - Dr Patrick Harrison, Department of Physiology at UCC and Professor Elena Levtchenko, Professor of Pediatrics, UZ Leuven.

The workshop programme itself comprised five keynote speakers with a further 15 speakers delivering short research communiques.

A particular focus of the Dublin Cystinosis Workshop 2020 was on the role of the patient voice both in directing their lives, their treatment, management of their disease and as partners in the research process – *'nothing about us, without us'*. In this context, there were important inputs from US contributors Ms Cheryl Simeons and Ms Karen Gledhill. Ms Simeons and Ms Gledhill, both of whom live with cystinosis, shared their experiences of being members of the Cystinosis Research Network's (CRN's) Adult Leadership Advisory Board which connects young adults with other peers to help empower each other by sharing stories and experiences and to maintain and strengthen their own disease management. Focusing on a different aspect of the patient voice, Ms Anne Marie O'Dowd

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emphasised the importance of patient experts input into the design, planning and execution of all research projects from basic science through to clinical trials

Dr Jennifer Hollywood won this year's Professor Roz Anderson Memorial Prize which was awarded for the best short communication presented at the workshop. Dr Hollywood presented her research on mTor inhibition in cystinotic isogenic pluripotent stem cells (iPSC) and the characterisation of an important new rat model to study cystinosis (which was supported in part by a Cystinosis Ireland Seedcorn Grant). Dr Hollywood was awarded the prize following a direct poll of her scientific peers participating in the workshop. Despite the significant time difference between Ireland and New Zealand, Dr Hollywood, participated during the whole workshop right throughout the night and was also a key member of this year's DCW scientific organising committee. We in Cystinosis Ireland are delighted that Dr Hollywood won this year's award and feel that she was a particularly worthy recipient not just for her excellent presentation but also for her dedication in participating all through the night right to the end of the workshop. The Professor Roz Anderson Memorial Prize is awarded in recognition of the immense scientific contributions to the field of cystinosis research made by the late Professor Roz Anderson, University of Sunderland. The Professor Roz Anderson Memorial Prize is €200 cash award and an invitation to present at the next annual Dublin Cystinosis Workshop.

Hosting the Virtual 6th Annual Dublin Cystinosis Workshop 2020 would not be possible without the hard work and assistance of very many people who regularly step up to the plate to help us out. In particular, Cystinosis Ireland would like to thank the members of the Dublin Cystinosis Workshop Scientific Organising Committee – Dr Paddy Harrison (Chairperson), Dr Atif Awan (Irish Medical Organiser), Professor Elena Levtchenko, Dr Achim Treumann, Dr Thomas J. McDonald and Ms Anne Marie O'Dowd who give their time and expertise freely and generously. Sincere thanks also to Ms Denise Dunne and Dr Ruth Davis for all their hard work in organising this event.

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