



THE CYSTINOSIS Advocate

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From Nashville to Virtual-ville

CRN Summer Conference Moves Online

Our biennial conference scheduled for this summer will still take place - virtually. With the safety and health of our community in mind, we've decided to shift from an in-person event to an online conference series. We are focused on providing an educational, supportive, engaging and fun experience. The beat will go on! Our original 2021 conference location of Nashville, Tennessee will now host our face-to-face celebration in 2023. Plus, we hope to host more in person regional gatherings in 2022.

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Please contact Jonathan Dicks at: **513.394.0681** | jdicks@cystinosis.org
553 Beechtree Drive, Cincinnati, OH 45224 or visit CRN.PCsRefurbished.com

This offer is available for individuals and families living with cystinosis. If you know someone who could benefit from this service, please pass it along.



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The President's Letter

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Maya Doyle, MSW, Ph.D., LCSW-R

2020 has seriously been one bizarre year. I don't think anyone would disagree. It started off like normal, but come spring the entire world changed almost overnight. A day by day cautious approach is the only way I can think to describe it. New rules, new restrictions, new worries and a search for a new normal overwhelmed most of us. But what were we to do? We had to stay safe, protect those we love and still somehow try to continue on with our lives. As if life isn't challenging enough right?

Most of us have found ways to deal with the changes and challenges this year has brought. I, myself, have found ways to be as safe and cautious as possible and still continue to operate my small business and occupy the few free hours I can find. The Cystinosis Research Network is no different. The volunteers at CRN have mostly always worked together on a "virtual" basis as it was, but still had to pull together to make some critical decisions on how to operate and provide to this community everything that we could, but in a safe and respectful way. Some parts of that were easy, while others took more thought and effort. Regardless of the amount of effort any project or undergoing takes, the CRN will continue to provide every resource possible. Through any pandemic and all of the things that life is throwing at us right now we are here and always will be.

Hopefully the President's Letter in the spring will have me bidding a huge and long awaited farewell to COVID and a huge welcome back to our normal lives.

Sincerely,

Clinton Moore



The Moore Family



Clinton Moore – masked up! Get yours at cystinosis.org/face-mask-fundraiser

Brighter Days Care Package Program

By Anna Pruitt

Brighter Days was created to give back directly to our cystinosis community through developing care packages. Our mission is to reach families going through challenging times, whether that be newly diagnosed, currently going through kidney transplantation or to remember and honor our loved ones that are no longer physically with us, has been such an honor.

Since launching in July 2020, Brighter Days has reached over 15 families. Brighter Days will continue to give back directly to our community in hopes of touching even more lives. We truly hope that we can brighten your day during the most challenging times of your cystinosis journey. We encourage families to stay tuned for more updates with Brighter Days. We hope to have additional ways of giving back to be announced in the near future.

To request a care package or learn more, click [here](#).



Attention:
Cystinosis Research
Network has a
new address!



Please send any inquiries, general correspondence and donations to:

The Cystinosis Research Network
P.O. Box 702
Lake Forest, IL 60045-9998

CRN Virtual Meetups: COVID-19 Creates a Need to Pivot

The Cystinosis Research Network holds biennial family conferences hosting many individuals, families and professionals impacted by cystinosis. In the years in between we've managed to bring together smaller groups in select cities around the U.S. Abiding by COVID-19 restrictions and keeping the health of our community in mind, this year those in-person conferences transformed into a series of virtual meetups.

Despite the "Zoom fatigue" that has set in for many, there were silver linings to be found in this transition. It afforded us the ability to engage with people outside of the United States and expanded the sessions to those who cannot travel, perhaps on dialysis, etc. We received over 70 registrants from across America, Australia, Canada, Mexico, New Zealand, UK, France and Germany.

Meetup topics focused on:

- **Mental Health** with Dr. Courtney Taylor Zimmerman
- **Research & Cystinosis Gene Therapy** with Dr. Paul Grimm and AVROBIO
- **Connecting Caregivers**
- **Cystinosis Basics** with Dr. Laurence Greenbaum

Excluding the private conversation Connecting Caregivers, recordings for each meetup can be found on the [CRN YouTube channel](#).

2020 **VIRTUAL MEETUPS**



Don't forget to visit the [events section](#) of our website for information on video conference calls led by the Adult Leadership Advisory Board where monthly conversations take place. This "Cystinosis Sessions" series is led by adults living with cystinosis.

Contribute to Your Community – ALAB Now Accepting Applications

We are thrilled to announce that ALAB (Adult Leadership Advisory Board) is extending new member

recruitment into the new year. During this difficult and isolating period, we want to ensure our volunteer

opportunity is available and accessible to all people around the world. If you are an adult (18+ years old) living with cystinosis, please consider joining efforts with ALAB to support the entire cystinosis community in 2021!

Learn more about ALAB's goals, current programs and how to [apply here](#).



Founding ALAB members at the 2019 CRN Conference



Virtual 6th Annual Dublin Cystinosis Workshop 2020

By Ruth Davis

After the incredibly successful Cystinosis Network Europe's Virtual International Cystinosis Conference which was held online in April 2020, Cystinosis Ireland was delighted to host the 6th Annual Dublin Cystinosis Workshop virtually on 16 July 2020.

The Dublin Cystinosis Workshop is a specialist scientific meeting aimed 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 its 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.

Mr. Mick Swift, Chairman of Cystinosis Ireland welcomed all of the participants to the meeting and reflected on the importance of maintaining links and the exchange of research advances during the COVID-19 pandemic. The virtual Zoom meeting was ably co-chaired by long time champions of Cystinosis Ireland - **Dr. Patrick Harrison**, Department of Physiology at University College Cork and **Professor Elena Levchenko**, Professor of Pediatrics, UZ Leuven in Belgium.

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

The keynote speakers were: (i) **Dr. Rik Gijssbers**, Associate Professor

in the Faculty of Medicine and Head of the Subdivision Viral Vector Technology and Gene Therapy, at KU Leuven, Belgium who provided a timely overview of the two types of viral vectors systems currently being used to deliver genetic material into a patient's cells; (ii) **Professor Michael Taggart**, Chair of Reproductive Sciences, at the Cardiovascular Research Centre, Newcastle University, UK, who delivered a very interesting presentation that considered the potential for cystinosis patients to develop cardiovascular problems in later life; (iii) **Professor Francesco Emma**, Head of the Department of Pediatric Subspecialties, Ospedale Pediatrico Bambino Gesù, Rome, Italy and (iv) **Dr. Manoe Janssen**, Assistant Professor, Department of Pharmaceutical Sciences, Utrecht University, both of whom gave

presentations on the search for new and more effective drug therapies for cystinosis; and (v) **Professor Maya Doyle** Associate Professor of Social Work, School of Health Sciences & Clinical Assistant Professor, Frank H. Netter MD School of Medicine, Quinnipiac University, USA and her co-contributors **Ms. Cheryl Simoens** and **Ms. Karen Gledhill**, who spoke about the need to hear the voice of the patient and their experiences establishing the Adult Leadership Advisory Board (now an important part of the Cystinosis Research Network), which is a forum for the cystinosis community to gain new perspectives and better navigate their unknown future by promoting honest and transparent dialogue about the challenges and negative aspects of the disease, while highlighting the positives.

Cystinosis Ireland was particularly grateful to Professor Emma for joining the workshop despite the immense workload pressures that he and his medical colleagues are experiencing in Italy due to the impact of COVID-19.

Short research communications were given by: (i) **Dr. Jennifer Hollywood**, from the University of Auckland who 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); (ii) **Dr. Shu-Dong Zhang**, principal investigator and senior lecturer at the Northern Ireland Centre for Stratified Medicine, University of Ulster, C-TRIC, Derry/Londonderry who presented his work on using gene

expression connectivity mapping to screen a database of over 1,000 FDA approved drugs in order to identify potential new therapies for cystinosis; (iii) **Dr. Koenraad Veys**, pediatrician and researcher at the Department of Pediatrics at UZ Leuven, Belgium who described a retrospective study on a cohort of cystinosis siblings to determine the impact of commencing cysteamine treatment early in the life of the patient on the progression kidney disease and other aspects of the cystinosis disease; (iv) **Dr. Aude Servais**, senior nephrologist at the Necker Hospital in Paris, France, also presented on RaDiCo-ECYSCO - a multi-centre European cystinosis retrospective and prospective cohort of cystinosis patients; (v) **Dr. Brendan Keating**, principal investigator at Penn Transplant Institute, University of Pennsylvania, USA who explained the plans of the iGeneTRaIN consortium to conduct genome wide genotyping and whole exome sequencing of all available cystinosis patients worldwide; (vi) **Dr. Swastika Sur** from the Sarwal Group, Department of Surgery, University of California San Francisco, USA, presented on her recent research findings on the potential role of the vacuolar ATPase gene family in kidney function and the male reproductive system in cystinosis, and (vii) **Professor Justine Bacchetta** from the Centre de Référence des Maladies Rénales Rares, Centre de Référence des Maladies Rares du Calcium et du Phosphore, at the Hôpital Femme Mère Enfant in Bron, and from the INSERM 1033 Research Unit in Lyon, France, who spoke about a research study CYSTEABONE which is focused on understanding the specific

genetic factors and therapeutic regimes and their impact on bones in cystinosis patients.

This year's Dublin Cystinosis Workshop also strongly encouraged early-stage researchers to present their latest research findings. Postgraduate research students, Dries David, KU Leuven; Amer Jamalpoor, Utrecht University; Fatima Tokhmafashan, McGill University; Annika Ewart, Hannover Medical School, and Meisha Khan, UCSD, all presented their current research at the workshop.

We particularly welcomed PhD student **Amer Jamalpoor**, the recipient of the first Professor Roz Anderson Memorial Prize for best scientific poster and also of the Cystinosis Ireland Prize for best lay-oriented oral presentation at the 5th Annual Dublin Cystinosis Workshop 2019, who was specifically invited to present on his research. Amer's presentation focused on investigating bicalutamide, a registered drug molecule, as a potential therapy for cystinosis. Amer demonstrated that bicalutamide had potential to correct aspects of kidney disease associated with nephropathic cystinosis when used in combination with cysteamine. The cysteamine-bicalutamide combination treatment was found to reduce cystine levels more than cysteamine alone, showing potential as a future therapeutic agent for cystinosis.¹

A particular focus of this year's Dublin Cystinosis Workshop was on the role of the patient voice both in directing their lives, their treatment, management of their disease and as partners in the

¹Jamalpoor, A. et al. (2020) Cysteamine-bicalutamide combination treatment restores alpha-ketoglutarate and corrects proximal tubule phenotype in cystinosis (preprint) doi: <https://doi.org/10.1101/2020.02.10.941799>

research process – ‘nothing about us, without us’. **Professor Maya Doyle**’s keynote presentation focused on the vital role that peer support plays amongst those living with chronic health conditions such as cystinosis as a means for improving knowledge and coping mechanisms, improving patient communication with healthcare providers, supporting better adherence with various drug regimes, reducing feelings of isolation, and most importantly, empowering individuals in their health decision-making and advocacy efforts. The Adult Leadership Advisory Board (ALAB), which was established by CRN in 2018, aims to nurture patients who have cystinosis, and provide guidance along the disease process with advice, support, and companionship. Professor Doyle’s co presenters **Ms. Cheryl Simoens** (ALAB Chairperson) and **Ms. Karen Gledhill** (founder member of ALAB) are both members of CRN’s ALAB. Ms Simoens and Ms Gledhill highlighted to the workshop the success of the ALAB to date and described the ALAB initiatives currently underway using social media communication technologies (video conferencing, podcasts and teen-oriented Instagram updates) to discuss issues such as mental health, relationships, drug regimen adherence and the various challenges and successes of members of the cystinosis community.

In addition to the keynote contributions by Professor Doyle, Ms. Simoens and Ms. Gledhill, **Ms. Anne Marie O’Dowd** Cystinosis Ireland Executive Member and Chair of the Worldwide Cystinosis Community Advisory Board, emphasised the importance of patient experts input into the design, planning and execution of all research projects from basic science through to

clinical trials. Ms O’Dowd emphasised to the audience that public and patient involvement (PPI) is no longer an optional extra when applying for research funding, it now influences whether or not a research proposal will be funded or not.

Dr Jennifer Hollywood wins the 2020 Professor Roz Anderson Memorial Prize for best short communication at the 6th Annual Dublin Cystinosis Workshop 2020

Cystinosis Ireland was proud and delighted to award Dr Jennifer Hollywood, Department of Molecular Medicine and Pathology, The University of Auckland, New Zealand, this year’s Professor Roz Anderson Memorial Prize 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.

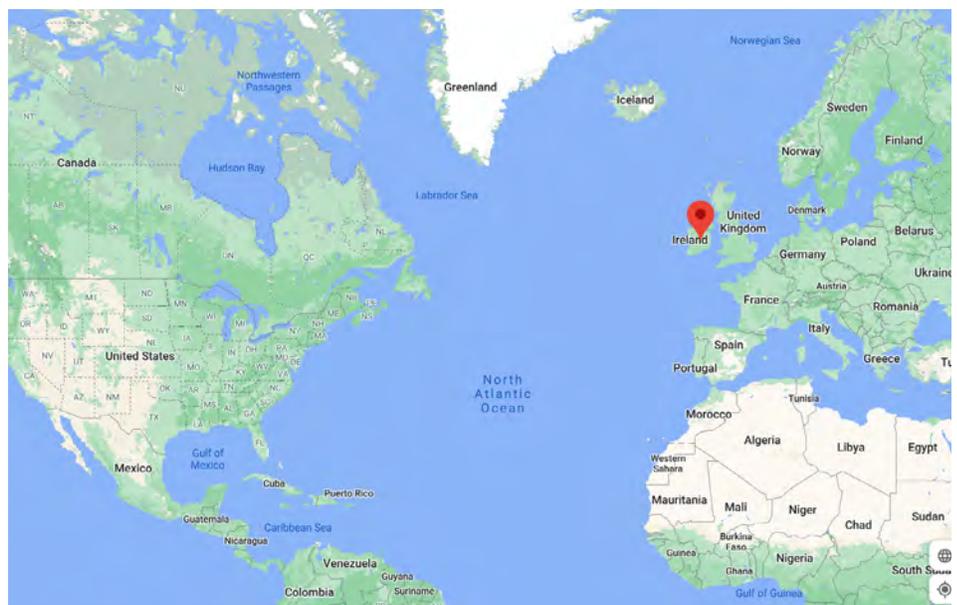
The Professor Roz Anderson Memorial Prize is awarded in recognition of the immense scientific contributions to the field of cystinosis research made by the late Professor Roz Anderson, University of Sunderland. The Professor Roz

Anderson Memorial Prize is €200 cash award and an invitation to present at the next annual Dublin Cystinosis Workshop.

Acknowledgements

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 Levchenko, 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.

A particular note of thanks must also go to Dr Harrison and Professor Levchenko, both of whom were fantastic chairpersons throughout the workshop, maintaining good order and promoting excellent discussions among the participants.



Mexico Updates

By Victor Gomez

A time together before COVID-19 hit the world. "Cystinosis Family Day" was held March 6-7, 2020 in Acapulco Mexico.

For the very first time our organization brought patients and families together at the beach! Not even COVID-19 was an obstacle at this year's family conference. Some of the highlights included:

- Leonardo Rojas, a patient with Fabry disease and member of the Mexican organization for Rare Diseases Patient Assistance, was our motivational speaker. He spoke about his story fighting with this rare disease and how he is doing great right now. Leonardo is a radio announcer at a Michoacan Mexico local radio station
- For the very first time we had a mental health expert, Marilu Narvaez in attendance. She is a psychologist and family therapist. We organized a family session which was based on team work and positive thoughts about our future dealing with a rare disease
- International Speaker Paul Grimm gave a cystinosis overview, research updates, recommendations for patients and answered questions from patients

The Mexican Organization of Cystinosis is committed to raising awareness and patient assistance. These conferences are very helpful in making it happen.



For years, the Mexican Organization for Cystinosis has been working to make cystinosis treatments available. Please ask your doctor if these medications may be an option for you and your family.



Thank you to all who attended!

Cystinosis Update from Egypt

By Neveen A. Soliman

All cystinosis heroes here are on regular cysteamine therapy and the cystinosis clinic has been renovated recently. Virtual clinics are also operational for some patients given the COVID-19 circumstances.

Neveen A. Soliman is Professor of Pediatrics & Pediatric Nephrology and Vice Dean for Research & Postgraduate Affairs, Kasr Al Ainy School of Medicine, Cairo University. She is the Founder and Director of the Egyptian Group for Orphan Renal Diseases (EGORD), the first group of its kind in Egypt and the region, to care for rare kidney diseases. She is also the founder of the National Association for Rare Diseases (NARD) and the African Inherited Kidney Diseases working group (AfrInKiD). Her

research interest is focused on rare, genetic and metabolic diseases particularly clinical and molecular characterization of cystinosis, ciliopathies, podocytopathies, primary hyperoxaluria, cystinuria, and collagen IV nephropathies among other inherited kidney diseases.

Professor Soliman is a member of several Pediatric Nephrology and Genetics scientific societies, she authored and co-authored numerous publications in renowned high-ranking journals and is regularly involved in pediatric nephrology education and training both regionally and internationally. She had received numerous honors and awards including, 2011 Global Kidney Academy/International Nephrology Education Foundation "Leadership

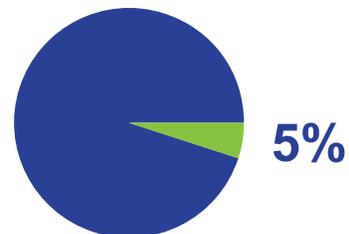


& Education in Nephrology" Award; 2012, 2015 & 2017 Cairo University Distinct Research Award; 2015 Prof. Ikram Abdelsalam Award in Medical Genetics and 2016 Scientific Research Academy Award.

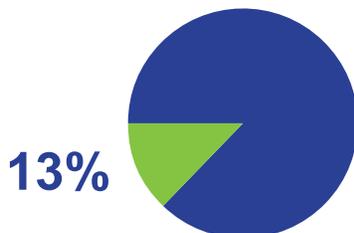
Cystinosis & COVID-19 Survey



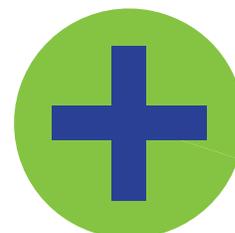
24 U.S. States responded & 9 non-U.S. respondents



5% experienced COVID-19 symptoms



13% received COVID-19 testing



2 responders tested positive for COVID-19

Based on these results, our medical advisory board feels that cystinosis patients are not affected any more frequently, or severely, than the general population. Results submitted through August 2020 and may not reflect current statistics.

Dutch and Flemish Cystinosis Group 20th Anniversary

By Marjolein Bos and Fons Sondag

Time flies. In 2000, Fons Sondag (father of a daughter with cystinosis) and Marjolein Bos (mother of a son with the same condition) both visited the first International Cystinosis Conference in Bergamo, Italy. Until then they had never met. Another Dutch family was present also and it was strange to visit Italy to meet Dutch families. Based on that fact the decision to start a Dutch group was made easily. In November 2000, the first Dutch meeting was held. Eighteen families responded to the invitation and nine families were able to attend in person.

During the past 20 years our group has grown to 50 patients and their families, also patients from the Flemish part of Belgium are welcome in our group. We are proud to say that young children who've visited our meetings together with their parents in the past still join us.

We organize annual meetings with medical and practical/daily life information. Every second year we have a family day with a children's program. It is so nice to meet each other's children and for the children to meet others with the same condition and also to meet as parents.

A lot has been changed during the last 20 years. Often people think that in the field of rare diseases nothing will change, but we are extremely happy that for cystinosis we have a dedicated community with key specialists and researchers.

TREATMENT

Cysteamine: Before 1997 cysteamine was provided as a food supplement,



the pharmacist made a liquid solution which was very unstable. We had to pick up a new bottle every week. In 1997, the Cystagon capsules were available on the market which was a huge step forward. But the six hour regiment of this medication is still a burden. Although our expertise center contributed to the trial, Procysbi is not reimbursed by our authorities. Until today our families and patients still suffer from the night dose of Cystagon.

Eye drops: In the past we had eye drops produced by the local pharmacist. The instruction for use of this product is every waking hour, one drop in each eye. This means for adult patients approximately 12 drops per eye a day. The questionnaire we sent to our members showed that 60 percent were able to use the eye drops 4-5 times a day, far less than

every waking hour.

For three years we've had the commercially available Cystadrops and, as a consequence, production by pharmacies is prohibited in the Netherlands. Unfortunately, the use of Cystadrops has side effects which influence the adherence to the therapy: the opening of the bottle is difficult for patients with muscle wasting. The dropper is stiff and not easy to handle. The drops are stinging in the eyes, causing a blurred view and crusts around the eye. We are in discussion with Recordati to solve these problems.

FERTILITY

A milestone in this 20 years was the research which showed that men with cystinosis can father a child. By harvesting precursor sperm cells and using an ICSI/IVF method, it was shown to be possible to induce a

pregnancy. Our group was extremely happy when, in 2014, the first twin was born from a father with cystinosis.

PATIENT CARE

The care for cystinosis patients improved a lot during the last 20 years. Professoer Elena Levtchenko started a clinic for adult patients with cystinosis with metabolic and nephrology doctors and taught them the specifics of cystinosis. This clinic also has an eye doctor, an orthopedic specialist and specialized nurses.

Dr. Marlies Cornelissen is head of the expertise center for cystinosis and manages the clinic for children with cystinosis at the Radboud university hospital in Nijmegen.

These clinics are members of ERKnet, the European Reference Network for rare kidney diseases.

SUPPORT AT HOME AND AT SCHOOL

The knowledge on support at home and in schools is exchanged during our meetings. Parents tell each other their own experiences. In the Netherlands and Belgium all kind of support is available, but the access to this support is often a challenge, due to all the parties involved in the funding (insurance companies, municipalities, central government).

INTERNATIONAL

Since 2019, the patient organizations in Europe organized themselves as Cystinosis Network Europe (CNE). The two main goals of this network are: organize the biennial International Cystinosis Conference and manage the worldwide Community Advisory Board (CAB). Next to the European countries also USA, Australia, South

Africa, Mexico, Turkey are members. The CAB contacts the pharmacists and researchers responsible for cystinosis medication to discuss new research and product development in order to get the patient voice heard.

BROCHURES

We started to write a list of tips and tricks (sorted for age) for food and medication intake. This is published on our website. Next to a general brochure with information about cystinosis for teachers, paramedical workers etc. we developed a brochure for general practitioners. This brochure has been translated into English, German and will be translated into French.

LOSS OF PATIENT MEMBERS

During the 20 years we lost 3 of our cystinosis patients. They will stay in our hearts forever.

Cystinosis Network Europe

By Denise Dunne



Cystinosis Network Europe (CNE) is continuing to meet on a regular basis to share information about events and opportunities internationally. We are delighted to have recently welcomed representatives from Mexico, Russia and Turkey to the organisation.

Following our virtual international conference in April 2020, CNE is proud to announce that our colleagues in the Dutch and Flemish Cystinosis Group will host the 2022 CNE International Conference. We are looking forward to another excellent meeting. The session recordings and translations of the 2020 meeting are

available online – www.cystinosis-europe.eu or www.cystinosis.ie.

The Worldwide Cystinosis Community Advisory Board (CAB) is continuing to meet to update and connect with researchers and sponsors of clinical trials. We are working towards virtual CAB meetings with a number of companies on topics of importance to the whole cystinosis community. If you would like any further information on the CAB, please get in touch with Denise Dunne (denise.dunne@cystinosis-europe.eu).

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Cystinosis Emergency Relief Fund



NORD[®]
National Organization
for Rare Disorders

What is the purpose of this program?

NORD's Cystinosis Emergency Relief Fund offers small grants to families or individuals diagnosed with Cystinosis and in need of financial support when faced with limited resources to pay for unexpected or emergency expenses.

Who is eligible to apply for NORD's Cystinosis Emergency Assistance grants?

This program is designed to help patients who:

- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address
- Have a diagnosis of Cystinosis
- Fall within the Program's financial guidelines and adhere to application requirements that are set in advance by NORD

What kinds of assistance can I request from NORD?

NORD's program can assist eligible individuals/families with two types of expenses:

- Unexpected or emergency expenses that an individual or family living paycheck-to-paycheck cannot afford without short-term assistance.
 - Some examples of these expenses may be the cost of diapers or repair costs for cars or major appliances, as well as travel expenses for clinic visits (lodging, gas cards).
- Monthly bills that an individual or family living paycheck-to-paycheck cannot afford to pay at the time of their application to NORD because of a documentable extenuating circumstance.
 - For example, if a parent or caregiver is required to take unpaid leave from work to care for an ill family member and cannot then afford the monthly cost of cell phones, or auto insurance, or rent/mortgage payments, then with these types of documentable emergency situations, NORD would consider making a one-time monthly payment to established bills with the opportunity to reapply if necessary and funds are available.

What is the application process?

Awards are granted on a first come, first served basis. Patients may be referred to the program by their health care provider, their case managers, or they may self-refer. The RareCareSM Patient Services Representative will guide the applicant through the application process during the initial phone call, at which time the representative will complete the Electronic Income Verification (EIV). If the tool identifies that the patient falls within the program's financial guidelines and the patient then agrees to NORD's Disclosure Agreement during a recorded telephone call, the application process will be concluded and the patient approved for assistance.

Frequently Asked Questions

What is NORD?

The National Organization for Rare Disorders (NORD), a 501(c)(3) organization, is an independent charity dedicated to the identification, treatment and cure of rare "orphan" diseases such as Cystinosis through education, advocacy, research and patient service programs.

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.

Funding for NORD comes from a variety of sources including corporate donations, foundation grants, public contributions, and membership dues.

Are there expenses which cannot be covered by NORD'S emergency assistance program?

Yes, NORD's goal is to be as flexible as possible in regards to patient's emergency needs, but some expenses which are not permissible, by law are:

- Federal, state, or local tax

How long before a decision is made on an application for assistance?

The application decision process can take as few as 5 minutes over the telephone. Applications completed and submitted via email, fax or US mail will be processed within three (3) business days of receipt.

What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The RareCareSM Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

Is there a limit to a patient's financial award?

A decision to place a "cap" on funding or limit the scope of assistance to beneficiaries is at NORD's discretion and is determined based on the amount of donations made to the fund, as well as the anticipated volume of applicants expected to utilize the program, and their anticipated financial need.

How do I apply for assistance from NORD's Cystinosis Emergency Assistance Program?

Phone: 855-201-5087

Fax: 203-486-8033

Email: cystinosis_assist@rarediseases.org

9am - 7pm (E.S.T.) Mon – Thurs and 9am - 6pm Fri

US MAIL to: NORD

Attention: Cystinosis Program

55 Kenosia Avenue, Danbury, CT 06810

payments, including property taxes, child support payments, legal fines and/or fees

- Luxury goods and services or vacation costs are not eligible for consideration
- Insurance premiums and copay out of pocket costs

Is there a fee for applying for assistance?

No, NORD does not charge our applicants when applying for help.

Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are offered as payment support for a one-time emergency need. Beneficiaries may reapply within that same year if funding continues to be available.

How does the payment or reimbursement process work?

NORD will either utilize a debit card allowing authorized purchases or will reimburse the patient directly for their approved expenses.

All claims submitted for reimbursement must be provided within 30 days and include receipts or other evidence of payment, such as a credit card statement.

Reimbursements will be made within ten (10) business days of receipt by NORD.



**Require financial help due to the pandemic?
COVID-19 qualifies as an unexpected emergency
under NORD's Relief Fund. Details on how/
where to apply is listed above.**



Education & Awareness

By Marybeth Kruppenacker, Vice President of Education and Awareness

As we begin to look ahead to 2021 and hope and pray that life can get back to what we consider “normal”, we have to look at 2020 and wonder... “what happened.” I think we are in a brave new world of advocacy and non-profit. CRN continues to be active and engaged with the community. While we have not been able to participate at “in-person” meetings and some of our traditional activities are certainly conducted differently...we remain quite busy as an organization.

We have traditionally attended the PAS (Pediatric Academic Society) meeting as well as ASN (American Society of Nephrology) and we were registered for both in 2020, but we do maintain our connections with them and monitor and update information about upcoming 2021 meetings. While some organizations have started to opt out of those “in-person” meetings, we would hope to resume attendance in the spring and fall of 2021. Stay tuned!

Our CRN mask project has been an overwhelming success! Thank you to the generosity of John Maccarone and Maccarone Plumbing and Heating and Richie Maccarone of Valley Sports, we were able to secure another 500 masks for our community. We have been blessed to be able to provide a free mask to each patient with cystinosis and charge \$10 for each additional mask. We have had requests internationally and mailed them from Canada to Australia! It has been a wonderful outreach to our cystinosis community and reaffirms that we are connected throughout the world! I have been blessed to hear from so many families and have loved seeing the photos posted of our families wearing their masks proudly!! Masks are still available on our [website](#) or by emailing mkrumm@aol.com.

One of the most wonderful things CRN continues to do is to provide academic scholarships to both our cystinosis patients and to siblings! With the help of our scholarship committee,

Gail Potts and Carol Hughes, we were blessed this year to provide six academic scholarships! In addition to our Individual Living with Cystinosis Scholarship and the Sierra Woodward Sibling Scholarship, we now have the Deanna Lynn Potts scholarship available to our community. Thanks Gail Potts and the generosity of her family. It is a wonderful feeling to read the thank you notes after these award notifications are made! It is genuinely appreciated by all recipients.

Our hope and our prayer as the next big event will be our 2021 CRN Family Conference, right now scheduled for the summer of 2021. While we are in the early stages of planning, we are constantly in contact with our venue and as we move forward with planning, we will keep the community informed for any updates! It truly is one of the best times we get to spend time together.

Family Support Update

By Jen Wyman, Vice President of Family Support

Living with, or caring for, someone with a rare disease naturally creates an uneasiness and fear of the future. Add a pandemic to that already magnified sense of fear and you may seriously consider wrapping your family in bubble wrap until it is over. It has been a time of heightened anxiety and alienation with stay at home orders and closures. While some have embraced the family time and “hunkering down” others have struggled with the stress of finances, health care, jobs, mental health. Each of us has experienced our own struggles and successes over this period. With social distancing measures in place, social media has become a place to stay in contact with one another and to maintain our sense of community. And while everything that comes across our feeds isn’t positive, healthy information or conversation it remains to be a platform that we can use to stay connected to each other.

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A community is a familiar thread used to bring people together to advocate and support each other in the fight to overcome those threats. As human beings, we need a sense of belonging, and that sense of belonging is what connects us to the many relationships we develop.

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We are fortunate to have an amazing cystinosis community given how small our numbers actually are. We are fortunate to have social platforms that keep us connected in times of unrest. We are fortunate to have each other.



CRN Facebook public page [@CystinosisResearch](https://www.facebook.com/CystinosisResearch) or [facebook.com/CystinosisResearch](https://www.facebook.com/CystinosisResearch)

CRN Facebook (closed/private) group [facebook.com/groups/6382741905](https://www.facebook.com/groups/6382741905)



Twitter
[@CystinosisCRN](https://twitter.com/CystinosisCRN)



Instagram [@cystinosisresearchnetwork](https://www.instagram.com/cystinosisresearchnetwork)

Instagram for teens (closed/private) [@CystinosisTEENS](https://www.instagram.com/CystinosisTEENS)



[Cystinosis Research Network](https://www.youtube.com/CystinosisResearchNetwork)



The Chodakowky Family

By Danielle Chodakowsky



Chase was born on February 16th, 2012 as a seemingly healthy baby boy weighing 8 lbs. 12 oz. His first year of life was what you would expect for a normal one year old. However, around 15 months old we started to suspect that something wasn't quite right. Chase would get very irritable and you could only console him with water and it seemed as though his thirst was never quenched. With the excessive thirst came excessive urination and he also started to lose interest in food. Over the course of three months we took him to multiple pediatricians only to be told that we were worrying too much and that he was just a thirsty boy. After demanding that they check his blood sugar, because we thought he had diabetes, a normal result was found and the doctor's once again told us to stop worrying. Another month passed and Chase was now losing weight, complaining of pain when he walked, and he just looked so weak and frail. At this point, we called the pediatrician and begged for them to run some tests on Chase. The pediatrician agreed to take a look at his urine and that's where things started to change. After having two urinalysis that showed significant findings we were finally referred to an endocrinologist. The endocrinologist

ruled out any hormonal conditions causing these symptoms and we were then sent to a pediatric nephrologist. Chase's nephrologist took one look at Chase's physical characteristics and his symptoms and said, "look at him, he is a cystinosis baby!" Fortunate for us, the nephrologist had experience with another cystinosis patient many years prior and she was able to recognize this ultra-rare disease. At that point, Chase was diagnosed with Fanconi syndrome and failure to thrive. After eight long days, the bloodwork confirmed that Chase had nephropathic cystinosis. Chase was 18 months old at diagnosis.

Within a month from the date of diagnosis, we made the decision to have a g-tube placed to be able to provide Chase with the food/nutrition that his body so desperately needed and to also be able to administer his growing number of medications to manage cystinosis. The first year was rough. The necessary medications made Chase so nauseous and it was not uncommon for him to vomit multiple times per day. Chase slowly regained his strength but the nausea and food aversions continued. Chase didn't eat much by mouth for the next few years, his g-tube was a life saver!

There have been other issues that have come along with cystinosis, including the diagnosis of Pseudotumor Cerebri. Chase has had six spinal taps over the past few years to drain the excessive spinal fluid that builds up with this condition. These must be done to help preserve his vision.

Chase is now eight years old in third grade and is doing so well! He has

grown so much and eats just about anything these days! We no longer need to use the g-tube for food but we do still use it for administering medication. We are working on it but Chase is still not able to swallow his pills yet. Chase gets medication six times a day which includes 22 pills, six liquid medications, and administers his own eye drops 10 times per day. On top of his medicine, Chase drinks close to two gallons of water a day to try to quench his never-ending thirst! With all of this, he never complains and always has the most contagious smile on his face. Chase truly is the sweetest, most caring and loving little boy you'll ever meet and we are so blessed to call him "ours!"



JoJo: A Special Gift

By Ashley Haynes

On October 2, 2020, Maggie Jo Haynes received the best possible gift. She received a new kidney. This gift came from a person she did not know just one week earlier.

Maggie Jo was diagnosed with cystinosis when she was 1 year old. Her sister, Lily Grace, had been dealing with health issues since she was an infant and, at 4 years old, she was finally correctly diagnosed with cystinosis. Since it is genetic, Maggie was tested. We were very surprised when the results showed that she had cystinosis, too. Maggie Jo just didn't seem sick. In fact, she had a very atypical experience with cystinosis as a child. She took all of her medications and went to the scheduled doctor visits and labs, but she had few of the complications and issues that most children with cystinosis have. She rarely had nausea or low energy. In fact, Maggie played softball for many years and is an avid hunter. She was active and otherwise healthy.

It was when Maggie was ten years old that she first had a complication that is believed to be related to cystinosis. She had to have a guided growth surgery to correct her being "knock-kneed." Within two years, Maggie Jo had three surgeries to correct her legs. This had taken its toll on Maggie's energy level. And then, about a year later, when she was 13, she was admitted to the children's hospital for dehydration. This was the first time that her labs showed an abnormality (other than the cystine level that helped with her diagnosis). After this hospitalization, Maggie's kidney function declined quickly for about a year. In February, Maggie's



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Over the next few weeks, our family heard from 10-15 people who wanted to begin the process to see if they qualified to be her kidney donor. We were amazed.

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doctor told her that she would need to be seen for an initial evaluation for a kidney transplant. The appointment was made for late March.

Unfortunately, COVID changed our plans. Her appointment had to be backed up from March to April and then from April to May. Meanwhile, her lab visits continued to show a rapid

decline in kidney function. We began to get very concerned.

When we finally went for Maggie's initial transplant evaluation, we discussed possible living donors and the process of getting added to the deceased donor list. I had given my kidney to Maggie's sister, Lily, in 2011. We had no family members who were



a blood match. My sister, Alyson, was willing to do a paired donor match, but that was our only family possibility. While Maggie was at her evaluation, it was found that some of her levels were “off” and she needed to be admitted to get those corrected. While we sat in her hospital room that night, I posted on Facebook about Maggie’s situation and gave information in case anyone was interested in being a living donor. Two minutes after I posted, I received a message from a friend of my sister. She asked for the contact information. Over the next few weeks, our family heard from 10-15 people who wanted to begin the process to see if they qualified to be her kidney donor. We were amazed. Now, I will say that we have experienced the kindness and generosity of our community in the

past. When Lily received her kidney transplant, our community rallied around us and we definitely felt the love. But, the thought of all of these people willing to undergo surgery and give up an organ for our sweet girl, was humbling, to say the least. Because of privacy laws, we were not able to be informed about how things were going with potential donors. But, my sister’s friend kept us informed about where she was in the process. In mid-August, we got the news that she was in the final stages of the process. In late August, we were told that Maggie Jo would need to begin dialysis unless a transplant was scheduled in the next few weeks. We weren’t sure if the timing was going to work out. Finally, in mid-September, a date was set. Maggie would be getting her new kidney on October 2nd!

Maggie wanted to meet the angel who was willing to give this incredible gift. On September 27th, Maggie met Joy. Yes, Joy. The person giving the gift of life has the most appropriate name possible. Joy has two children. Her daughter is the same age as Maggie. On the day we met, gifts were exchanged and pictures taken. One week later, one of Joy’s kidneys was giving Maggie a second chance. And Maggie named her new kidney, JoJo, for Joy + Maggie Jo. JoJo is already working hard and allowing Maggie to have the energy that a 14-year-old should have. We hope that Maggie and Joy’s story is an inspiration to others who might consider being a living donor. It has certainly changed our lives.

Shields Family Story

By Laura Shields

We are Jon and Laura Shields of Nashua, New Hampshire. Jon and I have been married just barely two years. Our son, Zeke, was born in January of this year (really the only good thing about this year, stinkin' COVID-19). This year has been absolutely crazy for everyone, but I feel like we win the award for craziest with this news. I'm sure you all can relate.

Despite all this year has brought, we feel so blessed. We have so many reasons to be grateful. Zeke was diagnosed very early at seven months old (we were told this is early anyway). We are grateful that his prognosis will be better due to catching it so early. We feel so blessed to be a part of the cystinosis community too. I mean, obviously the requirements

to be a part of this community suck, but we've made so many friends who have shown incredible kindness to us in this difficult time. We've been sent numerous packages with such helpful information and items to help Zeke. I'm grateful for the guidance we've received from this group too. I mean, we have excellent doctors in Boston, but they don't know all the things we need to care for our son. And that's another thing to be grateful for, that I live near such great medical care. It could've been weeks or months before we received a diagnosis, but we got a differential diagnosis in one day, and he was correct. Most of all, we're grateful for Zeke and his amazing attitude and acceptance of all of this. He takes all his meds orally and he does fantastic. He is a warrior and we couldn't be prouder.



Industry Partnering

By Jonathan Dicks, Vice President of Development



We are cultivating new partnerships and projects with our incredible pharmaceutical teams at Horizon Therapeutics, Leadiant Bioscience, Recordati and AVROBIO who all

share the same conviction that each person with a rare disease has the right to the best possible treatment. These partnerships pave the way for new grant funding, medication development, as well as pipeline projects that are soon to come to fruition, offering our community incredible and completely-free services related to technology, education and family support. I am overjoyed at the prospects that are currently being fleshed out.

One such pipeline project on the verge of completion is the partnership with PCs for People, a 501(c)(3) nonprofit

organization responsible since 1998 for the distribution of over 150,000 computers to eligible families. Their digital inclusion efforts are essential in today's society, where it is increasingly hard to access education, healthcare and employment opportunities without access to a computer and the internet. CRN is proud to announce we will soon have the resources to offer our families and patients in need; high-quality refurbished desktops, laptops, internet and accessories at absolutely no cost. For details, visit [CRN.PCsRefurbished.com](https://www.CRN.PCsRefurbished.com).

CRN Scholarships Awarded

By Gail Potts

This year, the Cystinosis Research Network is happy to announce that we had six applications for our academic scholarships. We were able to grant all our applicants with these allocated funds to enable them to further their secondary education.

The CRN is happy to congratulate the 2020 recipients of the Sierra Woodward Sibling Scholarship, Individual with Cystinosis, and Deanna Lynn Potts Scholarship Awards.

INDIVIDUAL WITH CYSTINOSIS



Victor Gardner

The recipient of the Individual with Cystinosis awards was Victor Gardner. Victor is attending Cleveland State University majoring in speech and hearing. Cystinosis has inspired him choose a health care profession because of the care he received from all the health care professionals over the years.

SIERRA WOODWARD SIBLING SCHOLARSHIPS

The Sierra Woodward Sibling Scholarship went to three siblings of individuals with cystinosis: Kole Binger, Sarah Roberts, and India Gardner.



Kole Binger (left)

Kole will attend the Medical University of Wisconsin having graduated from the University of Wisconsin. Being a sibling of an individual with cystinosis had a profound impact on her career path and she hopes to go into research as a physician.



Sarah Roberts

Sarah hopes to pursue a career in either the legal or political fields in order to be an advocate for individuals with disabilities at the University of Victoria in Canada. By being a sibling she recognizes the governmental inequalities of disabled and disadvantaged individuals in Canada and wants to be able to give them a voice.



India Gardner

India will be attending Bowling Green State University in Ohio, where she is pursuing a degree in broadcast communication or journalism. She hopes to work in television some day to become a newscaster or journalist where she can give a voice to individuals with rare illnesses.

DEANNA LYNN POTTS SCHOLARSHIPS

We were pleased to award two Deanna Lynn Potts Scholarships this year, which went to Hannah Creel and Mason Reed.



Hannah Creel

Hannah is attending Samford University in Alabama, where she is majoring in music. She contributes her ability to overcome her fear of failure to her choir director in high school. In doing so it has led her to her musical aspirations.



Mason Reed

Mason will be attending Texas Tech University in Lubbock, Texas. He has not chosen a field of study, but has many interests. He was on the golf team throughout high school, was a filmer for the football team and kept books for the basketball team. He was also on the accounting team and participated on the robotics team. He was

a competitor in 4-H from a young age, and was proud to show his award winning swine.

Our sincere best wishes to all our amazing scholarship winners for a successful future.

Join the Cystinosis Research Network

Get connected! Stay informed! Together we can find a cure!

Become more active within our global network of caring families, concerned individuals and healthcare professionals working together in the fight against cystinosis. The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis. The Cystinosis Research Network is a volunteer, non-profit organization dedicated to advocating and providing financial support for research, providing family assistance and educating the public and medical communities about cystinosis. CRN funds research and programs primarily through donations from the public, grassroots fundraising events and grants. CRN provides outreach and access to resources. We take great pride in carrying out our motto:

“Dedicated to a Cure. Committed to our Community”...whether you are...

- **A Parent** who needs critical resource information, support services or help in sharing the challenges of cystinosis to those who serve your child.
- **An Adult** with cystinosis interested in information regarding medical and social issues that are specifically geared for adults or contribute your voice to new and legacy programming.
- **A Relative or a Friend** who wants to increase their understanding of cystinosis and find out how you can help out or become involved.
- **A Physician, Social Worker, Educator or other Professional** who makes a difference in the life of a family affected by cystinosis, and want to have access to critical information to better serve your patient, student or client.



The Cystinosis Research Network is proud to provide valuable resources to the community, free of charge. Resources include but are not limited to:

- The latest cystinosis information through our biannual CRN Newsletter, our website (cystinosis.org), the popular online Cystinosis Facebook Support Groups, regular email updates and social media channels.
- CRN Family Conferences and Regional Meet Ups. Exchange knowledge and create friendships with other families and individuals living with cystinosis. Learn first-hand the latest discoveries about cystinosis from the medical professionals.
- Rare Disease Week Scholarships. Participate in a week-long event in Washington, D.C. Let your voice be heard by legislators and policymakers who need to know why cystinosis (and other rare diseases) are important issues to you.
- Access to Cystinosis Research Network's representatives in the areas that are most relevant at any given time to you or your loved one affected by Cystinosis.



We'd Like to Stay in Touch

To best serve the cystinosis community, we have been working on reorganizing and updating the information in our database. While it can be unsettling to share personal information, please be assured that we use this database for CRN purposes such as mailings and reach select groups with opportunities and important updates.

Your feedback is greatly appreciated!

Full name	Email
Relation to person with cystinosis or "myself"	Street address
Name of the person with cystinosis (if not yourself)?	City, State/Province
Date of birth (person with cystinosis)	Zip/Postal code
Phone number	Country

If you are completing on behalf of more than one person with cystinosis, please continue:

Full name	Email
Relation to person with cystinosis or "myself"	Street address
Name of the person with cystinosis (if not yourself)?	City, State/Province
Date of birth (person with cystinosis)	Zip/Postal code
Phone number	Country

Return via email by scanning/uploading photo of this completed form to:

Jen Wyman at JWyman@cystinosis.org

**or mail to: Cystinosis Research Network, Inc.
P.O. Box 702, Lake Forest, IL 60045-9998**

Fundraising Update

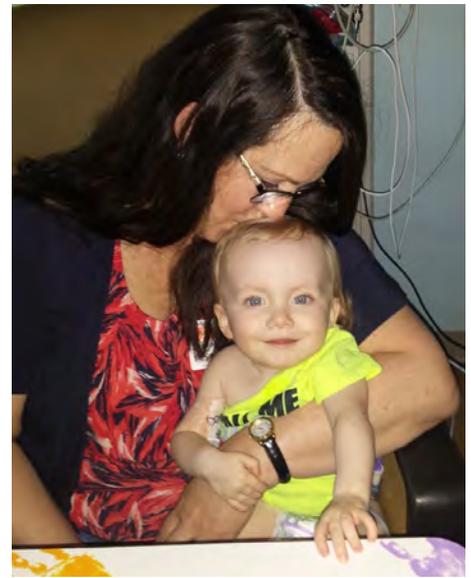


On the one year anniversary of Laura McGinnis' passing, the [Live Like Laura Fun Fund](#) (LLLFF) was launched. Laura lived life in pursuit of adventure, making memories every step of the way. In that same vein the goal is to enable people affected by cystinosis to participate in life's many adventures, paving the way to empower these warriors to create their own memories. The LLLFF helps to break down the financial barrier by offering up to \$1,000 to applying patients and siblings. To grow funding, we partnered with Frankie McGinnis to host a virtual 5k on November 1, 2020. Being virtual meant participants could be located anywhere in the world! A huge thank you goes out to all who made this inaugural event a success!



We also worked closely with the Peachman family bringing their annual golfing event Mulligans Fore Morgan to life during COVID! 2020 brought Morgan Peachman a new kidney, and a new lease on life. After turning 13 this summer, and hitting her six month kidney-versary, she was cleared for most normal activities. Morgan hit the ceremonial first tee shot! While nothing feels normal this year, The Peachman Family kept with the theme of changing times by bringing their fundraising event to TopGolf in Cleveland to ensure social distancing!

We are live with fundraising for the upcoming first annual 'Miles for Moose - 57 Miles for a Cure' slated for a May 8th, 2021 launch to acknowledge Cystinosis Awareness Day and the daily challenges little Benson (also as known as Moose) bravely faces each day. The goal of raising \$5,700 is already at 20% funded and continues to rise weekly! Please consider cross promoting, donating, or attending the 57 mile relay walk in Watertown, NY. Details can be found [here](#) and/or email Andrea to participate at milesformoose57@gmail.com. [See page 25](#) to learn more about Benson and the woman behind Miles for Moose.



Miles for Moose

By Andrea Carr

Our family lives in upstate New York. Some say the tundra of the Canadian border; others say the beauty of the shores of Lake Ontario, I think it all depends on the time of year. We are an unconventional family of marriages and separations, but we have two very special people that hold us all together, and that would be Benson, whom I affectionately call “Moose” and his sister Lillian who I also have a nickname for, “Bug.”

Benson was diagnosed with cystinosis in the spring of 2018. And while the diagnosis was absolutely devastating to him, his parents and our entire family, he is a true warrior and one that we all absolutely adore. Being only three, he of course doesn't truly understand his impact on those around him. His main focus right now is monster trucks, Mickey Mouse and how to torment his siblings. But we all see how much he has persevered since his diagnosis over two years ago.

Everyone in the cystinosis community knows how hard the initial diagnosis is.



“

What will it be like from this point forward?

What do you expect from day to day?

What is your new normal?

”

Some of you may not remember it as you may have been a baby, much like Benson was, when you were diagnosed, but your parents sure do, and your grandparents (like myself) and other family members do as well. And if they're anything like me, they felt absolutely helpless. Your parents are superheroes, literally. I commend every single one of you. But as a grandparent, all I could do was be supportive, and loving. So, I started thinking about fundraising, but not being familiar with it, I was a little lost.

One day, I was scrolling through my Facebook page, and a post from the Cystinosis Research Network popped up and it was about a man and his second annual 57-mile walk for Cystinosis Awareness Day on May 7th each year and for his son, Chandler. This man spent the first year of his event walking 57 miles by himself to raise awareness, and the second year, numerous people from his community joined to walk with him. Being incredibly inspired by him, my wheels began turning. I contacted Benson's mommy Larissa,

Aunties Paige and Mandie, Grandma Tami and Godparents Jasmine and Nick Sprague, and asked them if they wanted to be a part of a committee and pitched my idea... a 57-mile RELAY walk to bring awareness and to raise funds for the Cystinosis Research Network. Little did I know, the man in the original Facebook post was none other than Clinton Moore, the President of CRN. And whenever I reached out to him to find out just exactly how his walk developed, and to see if he had any great advice to lend, I also found a new team member for our Miles for Moose relay walk and a new friend.

Miles for Moose was created based off of Clinton's original remarkable idea, but it has grown to become so much more than I ever expected. While we always wanted the day of the walk to be a family friendly event, our community has become unbelievably involved as well. The original kickoff date was May 9th, 2020, but unfortunately due to our year of COVID, Miles for Moose's 1st Annual Relay Walk was cancelled. I have to admit, after so much hard work and planning, it was absolutely heartbreaking to postpone. But, ultimately it has become a blessing. We just held our second can and bottle drive with a bake sale included and raised almost \$1,800! While this year has been so tumultuous for many, I have tried to find the



(Left to right) Lillian, Andrea and Benson

“ Always remember you are **braver** than you believe, **stronger** than you seem, **smarter** than you think, and **loved** more than you know. – Winnie the Pooh ”

silver linings where I can, and I am determined to make our ACTUAL first year on May 8th, 2021 be even better! Through the planning of Miles for Moose, I have come to learn so much more about the cystinosis community and just how incredible it really is. I am so fortunate to have been offered a position on the Board of Directors. I can only hope being on the board

will not only help me learn more about cystinosis, the treatments available, the research, etc., but also bring me closer to the families that I have grown to admire and respect so much. I am so amazed by the cystinosis community, and immensely humbled by our local community for their constant support for Benson and our family.



Save the Date
Rare Disease Day
February 28, 2021

Research Update

By Christy Greeley, Vice President of Research



Alex, Christy and Jack Greeley

CRN has funded over \$4.5 million total in research grants and fellowship, including a cystinosis fellowship at the National Institutes of Health, research and education programs in the United States and many countries around the world including Egypt, Mexico, England, Scotland, Italy, Belgium, France, and much more. CRN has also co-funded research projects with Cystinosis Ireland. CRN research topics have focused on every aspect of cystinosis with the purpose of understanding the disease and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, fertility, improved medications, psychological and much more.

Call for Research Proposals 2020

The Cystinosis Research Network announced our Call for Research Proposals earlier this year. CRN

utilizes a Scientific Review Board comprised of leading experts on the disease of cystinosis which reviews grant proposals and submits funding recommendations to the Cystinosis Research Network. More specifically, the Scientific Review Board provides independent, objective review and recommendations regarding each research proposal utilizing grant review guidelines established by the Cystinosis Research Network and in accordance with the mission of the organization. Priority is given to interventional research, both clinical and basic, that will lead to improved treatments for cystinosis. New investigators are particularly encouraged to apply. The Chairperson of the Scientific Review Board summarizes its recommendations and presents them to the Cystinosis Research Network which then votes on each proposed project. A major focus of the Cystinosis Research Network continues to be a determined

effort to secure a promising future for the cystinosis community through the support and funding of research grants that lead to improved treatments and ultimately a cure for cystinosis.

2020 CRN Research Grant Funding

As a result of the 2020 Call for Research Proposals, CRN is proud to announce funding for the following research projects for a total of \$436,193 in new research grants.

Cognitive Control Systems in Cystinosis

Sophie Molholm, PhD Co-Principal Investigator, John Foxe, PhD Co-Principal Investigator

Grant Amount: \$315,193, two year study

Seminal behavioral studies by Trauner and others in human patients have suggested the presence

of cognitive dysfunction in cystinosis⁷⁻¹⁵. Our own work suggests some behavioral and neural differences

and difficulties with sensory memory in this population. However, the neurocognitive phenotype associated with CTNS mutations and its developmental path are still poorly understood, and the prime areas of neurocognitive vulnerability in this population are in need of much more thorough characterization. This is critical to developing effective therapies to compensate for or improve on areas of cognitive vulnerability. To this end, we propose to characterize different components of executive functioning (memory updating, set shifting, conflict monitoring, and inhibition)

in cystinosis. Focus on this area is motivated by our previous work, the literature on cognitive weaknesses in cystinosis thus far, and by first person reports collected during interactions with patients and families. We will use high-density electrophysiology (EEG)—a non-invasive method that allows one to directly measure functional brain activity at the millisecond scale and thus reliably assess the integrity of information processing at the neural level—and standardized cognitive functional assessments to test 15 children, 15 adolescents, and 15 adults with cystinosis and the same number of age-matched unaffected healthy controls. Twenty heterozygotes and age matched controls will also be tested to examine the role that mutation versus disease plays in the cognitive phenotype of cystinosis, Significance: Greater knowledge of the neurocognitive dynamics of executive function in cystinosis has the potential to critically advance the development of interventions for these individuals. In addition, identification of neural markers will provide objective assays of treatment efficacy on brain function.

Chitotriosidase as a therapeutic monitor for cysteamine therapy in cystinosis: a retrospective validation study

Mohamed A. Elmonem, Koenraad R.P. Veys, Lambertus P. van den Heuvel, William A. Gahl, Elena Levtchenko

Grant amount: \$44,000, one year study

Nephropathic cystinosis lacks a practical therapeutic monitor for cysteamine therapy. The currently used white blood cells (WBC) cystine assay, although very specific for the disease, has many clinical and technical drawbacks limiting its routine

use in most countries. Chitotriosidase enzyme activity in plasma is a potential alternative to WBC cystine in nephropathic cystinosis monitoring. Chitotriosidase is mainly produced in various tissues by activated macrophages upon stimulation by cystine crystals. We have previously demonstrated that the enzyme is significantly elevated in the plasma of cystinosis patients compared to both healthy and renal controls. Moreover, in a two-year longitudinal study chitotriosidase enzyme activity was a significant independent predictor of WBC cystine levels and was superior to WBC cystine as an indicator of the number of extrarenal complications in cystinosis patients. When compared to WBC cystine, plasma chitotriosidase assay is simpler, faster, more economical and needs a much smaller sample making it more convenient, especially in young children. Furthermore, the enzyme is extremely stable for years at minus zero temperatures. In the current proposal, we plan to validate our results in a large cohort of previously recruited cystinosis patients followed up at the NIH Clinical Center in Bethesda, Maryland, USA. Stored samples at -80°C of 150 cystinosis patients will be retrospectively assayed for chitotriosidase enzyme activity, and will be correlated to documented parameters of kidney function, extra renal complications and a WBC cystine-based long-term compliance score.

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis

Katharina Hohenfellner, MD

Grant Amount: \$77,000, one year study

The concept presented here, follows the established approach for the rehabilitation of children “Auf die

Beine”, which was developed in Cologne for children and adolescents with limited mobility, e.g. caused by cerebral palsy or patients with osteogenesis imperfecta (<https://unireha.uk-koeln.de/kinderjugendreha/behandlungskonzept-auf-die-beine/>) (3). The training design with home exercise and short training sessions considers the fact that the target patient group is already under great strain due to the severity of the disease (medication, special diet, possibly dialysis). The project follows the official recommendations for the use of a vibration plate (4,5). Patients train with Galileo vibration plates according to a fixed training schedule which provides for 10 short training sessions per week (maximum 2 per day). Within one training session, four exercises will be performed. The control group will perform the same exercises without vibration plates, but with dumbbells. Patients will initially undergo an intensive training course and will receive regular supervision during the three-month home training phase. The study is designed as a randomized controlled trial. Due to the very rare underlying disease a matched pair design was chosen to achieve comparability between the two groups. The patients will be matched based on their age, sex, and major previous surgeries. A baseline and two follow-up clinical assessment, one after the three-month home training phase and one after the follow-up phase, will take place. The primary endpoint is the change in muscle strength (in %) from the baseline examination to the measurement after the training phase. In the control group (CG), a mean shift of 0 % is expected, since no vibration training is carried out.

Potential Impact for patients with Cystinosis:

Patient orientation: empowering patients by supporting an active lifestyle and enabling patients to positively influence the course of the disease themselves. Optimization of clinical outcomes: improving cardiorespiratory performance and increasing muscle strength in patients. Improvement of patient-oriented end points of care: improving quality of life.

CRN and Cystinosis Ireland Co-Fund UCSF Study of Male Infertility

We are pleased to announce a collaboration between the Cystinosis Research Network (CRN) and Cystinosis Ireland to fund a male infertility in cystinosis study at the University of California San Francisco (UCSF). The collaboration was made possible, in part, from a private contribution from CRN board members, Anna and Paul Pruitt and from Cystinosis Ireland's Seedcorn Funding Programme. Dr. Sur, a postdoctoral fellow in Professor Minnie Sarwal's Laboratory at UCSF, is the Principal Investigator in the "Cellular Resource for Studying Male Infertility in Cystinosis" proposal. Both organizations look forward to this partnership and providing greater insights concerning cystinosis and male infertility.

This grant was awarded in July 2020 by the Cystinosis Research Network and Cystinosis Ireland.

A Cellular Resource for Studying Male Infertility in Cystinosis, Minnie Sarwal, MD, PhD, Professor of Surgery, (Director, Precision Transplant Medicine) University of California San Francisco (UCSF), James Smith, MD, MS (Director, Male Reproductive Health Center Urologist), University of California San Francisco (UCSF), Ann Harris, Professor, Department of Genetics and Genome Sciences, School of Medicine, Case Western

Reserve University, Cleveland, Ohio, Elena Levchenko, Professor, Department of Pediatric Nephrology, Leuven, EU, and Swastika Sur, MSc., PhD Postdoctoral Scholar, Sarwal Lab, University of California San Francisco, Department of Surgery.

Total Grant: €10,000

Principal Investigator, Swastika Sur, a Postdoctoral Fellow at the University of California San Francisco, has outlined a research proposal focused on infertility in men with cystinosis.

In addition to various endocrine organs that are affected in cystinosis, hypergonadotropic hypogonadism has been reported as a frequent finding in male cystinosis patients. Although spermatogenesis has shown to be intact at the testicular level in some patients, no male cystinosis patient is known to have naturally fathered a child. The sole treatment for cystinosis is the aminothioliol cysteamine, which is highly effective in reducing the intracellular levels of cystine. However, this drug is ineffective in the treatment of male infertility in these patients

A previous study of the pathophysiology of cystinosis-mediated infertility used a CTNS^{-/-} mouse model. However, the CTNS^{-/-} mouse model generated on C57BL/6 background was found to not be suitable for clarifying the pathogenesis of male infertility in cystinosis. This mouse model partially mimics the renal phenotype of the human disease but was found to have normal testicular morphology and function.

Therefore to this date, the exact pathophysiology of male infertility observed in patients with cystinosis is not yet fully understood, which is a critical unmet need due to the growing population of cystinosis patients, treated with cysteamine reaching

young adulthood.

This project proposes to generate isogenic immortalized epididymis and testicular cell models to study infertility associated with cystinosis, by using CRISPR/Cas9 technology to develop a deletion in the CTNS gene of normal human epididymis and testicular cells. The Sarwal group's ongoing collaborations with Dr Smith at UCSF has enabled access to a rich tissue resource of normal testicular and epididymal samples that will be used for generating this cystinosis-specific resource. The Sarwal research group also has ethical approvals in place at UCSF and at KU Leuven that will allow Dr Sur to approach cystinotic patients for access to patient biosamples.

Previously, the Sarwal research group successfully generated human immortalized CTNS^{-/-} proximal tubular epithelial cells, and confirmed the phenotype of these newly developed cell line in terms of intracellular cystine levels by HPLC-MS/MS.

In this project, Dr Sur will focus on generating human immortalized CTNS^{-/-} epididymal and testicular cells, followed by phenotype validation so that these cell lines can serve as a resource for the research community to study the pathophysiology of male infertility observed in cystinosis.

The Specific Aims of this project are the following:

Aim 1: Generate human immortalized CTNS^{-/-} epididymal and testis cell lines by CRISPR/Cas9 and confirm the phenotype to further downstream study of male fertility associated with cystinosis

Aim 2: Map the molecular perturbations in both cell lines with deletion of CTNS and in tissue samples from male cystinotic patients, by using state of the art genomics that

the Sarwal Lab has legacy expertise in. This will define the clinical utility of the resource generated in Aim 1.

This project will provide a first of its kind human cellular models to study cystinosis-mediated male infertility.

Cystinosis Community Advisory Board

I continue to participate as the U.S. representative in the Cystinosis EuroCAB programme, a project of EURORDIS, the European Rare Disorder Organization. The Community Advisory Board's (CAB's) objective is to improve patient access to novel therapies and treatments. This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research. We look forward to partnering with researchers and industry worldwide to improve the quality and speed with which cystinosis treatments are developed with the patient's voice in mind.

National Institutes of Health

As a reminder, patients may contact the National Institutes of Health to be enrolled in the cystinosis protocol and for consultative care. For more information, please contact:

Joy Bryant, (301) 443-8690,
bryantjo@mail.cc.nih.gov

Educational Resources

All of CRN's educational materials including brochures, guides and other publications have been updated and are available on the CRN Website. Look for a newly expanded Dialysis and Transplant section will include

a broad range of information and resources for those facing these challenges (cystinosis.org/about-cystinosis/dialysis-transplant).

Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from around the world. Also be sure to check out the many cystinosis related articles and publications available in our [Publications and Guides](#) library.

AVROBIO Announces New Patients Dosed in Gaucher Disease and Cystinosis Clinical Trials

First patient dosed in AVROBIO's global Phase 1/2 clinical trial of AVR-RD-02 for Gaucher disease type 1

Second patient dosed in investigator-sponsored Phase 1/2 clinical trial of AVR-RD-04 for cystinosis

CAMBRIDGE, Mass.--(BUSINESS WIRE)--Jul. 6, 2020-- AVROBIO, Inc. (Nasdaq: AVRO), a leading clinical-stage gene therapy company with a mission to free people from a lifetime of genetic disease, today announced that the first patient has been dosed in the company's GuardOne clinical trial, a Phase 1/2 investigational study evaluating AVR-RD-02 for Gaucher disease type 1. The company also announced that the second patient has been dosed in the ongoing investigator-sponsored Phase 1/2 clinical trial of AVR-RD-04 for cystinosis.

"The first patient dosed is an important milestone for the Gaucher disease community and our AVR-RD-02 program. Gaucher disease type 1 leads to an array of serious symptoms and the current standard of care does not halt disease progression," said Geoff MacKay, AVROBIO's president and CEO. "With a single dose of our investigational lentiviral gene therapy, we aim to prevent the buildup of a

fatty substrate in specialist immune cells called macrophages, as well as debilitating symptoms throughout the body, including the brain."

The company's Phase 1/2 trial of AVR-RD-02 for Gaucher disease type 1 is currently recruiting patients in Australia and Canada, with new clinical sites expected to open in the U.S. and Israel by year-end.

Gaucher disease is a rare, inherited lysosomal storage disorder characterized by the toxic accumulation of glucosylceramide (GlcCer) and glucosylsphingosine (GlcSph) in macrophages. Macrophages bloated with these fatty substances are called Gaucher cells which amass primarily in the spleen, liver and bone marrow. This results in a variety of potential symptoms, including grossly enlarged liver and spleen, bone issues, fatigue, low hemoglobin levels and platelet counts and an adjusted lifetime relative risk of developing Parkinson's disease that may be more than 20 times greater than the general population. Even on enzyme replacement therapy (ERT) – the current standard of care – people with Gaucher disease type 1 have a shortened life expectancy and may experience debilitating symptoms that significantly reduce their quality of life. An estimated 1 in 44,000 people are diagnosed with Gaucher disease.

"While the current treatments for Gaucher disease -- enzyme replacement therapy and substrate reduction therapy -- have been life changing, many unmet needs remain that significantly impact the daily lives of patients and families living with Gaucher disease, including fatigue, severe bone pain, joint destruction, increased risk of developing Parkinson's disease and other co-morbidities," said Christine White, executive director, National Gaucher

Foundation of Canada. “We welcome clinical trials of new therapeutics that have the potential to stop the progression of Gaucher disease and are excited to learn more about the potential use of this lentiviral gene therapy.”

The Phase 1/2 trial of AVR-RD-02 for Gaucher disease type 1 is designed to evaluate the safety and efficacy of the investigational gene therapy and is expected to enroll eight to 16 patients between the ages of 18 and 35. AVR-RD-02 starts with the patient’s own hematopoietic stem cells, which are genetically modified to express functional glucocerebrosidase (GCase), the enzyme that is deficient in Gaucher disease. The trial will include both patients who are treatment-naïve and who are on ERT. Every patient in this trial will be treated using the plato® gene therapy platform, AVROBIO’s foundation designed to scale gene therapy worldwide.

Second patient dosed in cystinosis clinical trial

The second patient has been dosed in the company’s AVR-RD-04 investigational gene therapy program for cystinosis. The ongoing Phase 1/2 clinical trial is sponsored by the company’s academic collaborators at the University of California, San Diego (UCSD)¹ and is led by Stephanie Cherqui, Ph.D., associate professor of pediatrics at UCSD.

Cystinosis is a progressive disease marked by the accumulation of cystine in cellular organelles known as lysosomes. This buildup can cause debilitating symptoms including kidney failure, corneal damage and thyroid dysfunction, often leading to a shortened lifespan. Currently, more than 90 percent of treated cystinosis patients require a kidney transplant in

the second or third decade of life. The current standard of care for cystinosis is cysteamine, a burdensome treatment regimen that can require dozens of pills per day and may not prevent overall progression of the disease.

The Phase 1/2 clinical trial is evaluating the safety and efficacy of AVR-RD-04 in patients at least 18 years of age who are currently being treated with cysteamine. The trial will enroll up to six patients. AVR-RD-04 starts with the patient’s own hematopoietic stem cells, which are genetically modified to produce functional cystinosin, the protein that is deficient in cystinosis.

Patient recruitment activities for Fabry Phase 2 trial ongoing

Patient recruitment activities for AVROBIO’s Phase 2 FAB-201 trial for Fabry disease continue for clinical trial sites in Australia, Canada and the U.S. While clinical trial sites are starting to reopen and patient identification activities are ongoing with a number of potential new patients identified, activities related to new patient screening, consent and enrollment in the FAB-201 clinical trial have been slowed because of the COVID-19 pandemic.

AVROBIO is conducting two clinical trials for its AVR-RD-01 investigational gene therapy for Fabry disease. Four patients have been dosed in the global Phase 2 trial (FAB-201), which is evaluating treatment-naïve patients, and five patients are participating in the fully enrolled Phase 1 investigator-led clinical trial, known as FACTs.

AVR-RD-01 starts with the patient’s own hematopoietic stem cells, which are genetically modified to produce functional alpha-galactosidase A, the enzyme that is deficient in Fabry disease. People with the

disease experience a toxic buildup of a complex cell lipid called globotriaosylceramide (Gb3 or GL3), which can damage tissues throughout the body and brain, and cause the progressive signs and symptoms of Fabry disease.

About AVROBIO’s personalized gene therapy approach

Our investigational lentiviral gene therapies start with the patient’s own hematopoietic stem cells. We use a lentiviral vector to transduce those cells in order to insert a therapeutic gene designed to enable the patient to produce a supply of the functional protein they lack. These cells are then infused back into the patient, where they are expected to engraft in the bone marrow and produce generations of daughter cells, each containing a copy or copies of the therapeutic gene. To optimize engraftment, we use a personalized conditioning regimen with precision dosing of busulfan to make space and enable durable engraftment in the patient’s bone marrow and central nervous system (CNS). Busulfan is an extensively validated conditioning agent generally considered to be the gold standard for ex vivo lentiviral gene therapy and has been administered to hundreds of patients for this purpose. Our approach is designed to drive durable production of the functional protein throughout the patient’s body, thereby potentially addressing symptoms from “head to toe,” including those originating in the CNS.

About lentiviral gene therapy

Lentiviral vectors are differentiated from other delivery mechanisms because of their large cargo capacity and their ability to integrate the therapeutic gene directly into the patient’s chromosomes. This



Christy Greeley with CRN face covering

integration is designed to maintain the therapeutic gene's presence as the patient's cells divide, which potentially enables dosing of pediatric patients, whose cells divide rapidly as they grow. Because the therapeutic gene is integrated using the vector into patients' own stem cells, patients are not excluded from receiving the investigational therapy due to pre-existing antibodies to the viral vector.

About AVROBIO

Our vision is to bring personalized gene therapy to the world. We aim to halt or reverse disease throughout the body by driving durable expression of functional protein, even in hard-to-reach tissues and organs including the brain, muscle and bone. Our clinical-stage programs include Fabry disease, Gaucher disease and cystinosis and we also are advancing a preclinical program in Pompe disease. AVROBIO is powered by

the plato® gene therapy platform, our foundation designed to scale gene therapy worldwide. We are headquartered in Cambridge, Mass., with an office in Toronto, Ontario. For additional information, visit avrobio.com, and follow us on Twitter and LinkedIn.

Collaborator-sponsored Phase 1/2 clinical trial of AVR-RD-04 is funded in part by grants to UCSD from the California Institute for Regenerative Medicine (CIRM), Cystinosis Research Foundation (CRF) and National Institutes of Health (NIH).

The full press release including AVROBIO's Forwarding-Looking Statement can be found [online](#).

The Cystinosis Research Network, Inc. Financial Review — Accrual Basis

By Jenni Sexstone, Treasurer

For the 9 months ending
September 30, 2020

Revenues

Total income for the nine months ending September 30, 2020, was \$440,000 compared to \$337,000 in 2019 due to increased fundraising efforts as well as corporate partner support.

Expenses

Total expenses for the period were \$136,000 compared to the same

period in 2019 of \$510,000. Education and conference spending was \$58,000 compared to \$353,000 in the prior year due to the family conference that took place in 2019. Year-to-date research grant expenditures were \$34,000 compared to the same period in 2019 of \$107,000. Total operating expenses of \$39,000 were slightly higher than the same period in 2019 of \$45,000 due to website and database updates that took place last year.

CRN had net operating income of \$304,000 for the nine months ending September 30, 2020. Continuous fundraising activities and generous corporate support continues to provide cash resources to increase patient advocacy and family support activities in 2020 and beyond to support the cystinosis community.

Cash on hand at September 30, 2020 was \$478,000. Net change in cash through the third quarter of 2020 was an increase of \$283,000.



Recordati Rare Diseases News Release

Recordati Rare Diseases Announces Availability of CYSTADROPS® (Cysteamine Ophthalmic Solution) 0.37% in the U.S.

Lebanon, NJ, September 15, 2020 – Recordati Rare Diseases Inc., today announced that CYSTADROPS® (cysteamine ophthalmic solution) 0.37% is now available for prescription and distribution across the United States (U.S.). CYSTADROPS is a new, viscous eye drop solution that depletes cystine crystal deposits in the cornea of the eyes of people living with cystinosis. Cystinosis is a rare genetic condition present from birth that leads to the build-up of cystine crystals throughout the body, causing widespread tissue and organ damage and significant impact on the eyes.

CYSTADROPS is available exclusively through Anovo, a specialty pharmacy that focuses on providing medical treatments to people living with rare and chronic diseases. Recordati and Anovo have partnered together to establish patient support services including insurance benefit investigations, educational references, resources for financial aid, and more. Clinicians can complete a prescription form [here](#).

CYSTADROPS is the first and only U.S. Food and Drug Administration (FDA) approved cysteamine eye drop formulation with four times a day dosing. The FDA approval of CYSTADROPS was supported by data from two clinical trials, both in which patients received CYSTADROPS at a median frequency of four times per day. A Phase 3 open-label, randomized, controlled, two-arm multicenter trial, with 15 patients in the CYSTADROPS arm, investigated the reduction in corneal cystine crystal density as assessed by in vivo confocal microscopy (IVCM). In the CYSTADROPS arm, the trial showed a 40 percent reduction in the IVCM total score across all corneal layers from baseline to 90 days.

A Phase 1/2a open-label, adaptive dose-response clinical trial of eight cystinosis patients showed that treatment with CYSTADROPS resulted in a 30 percent decrease in IVCM total score that was maintained for the five-year study period.

The safety of CYSTADROPS was evaluated in two clinical trials. The most commonly observed adverse reactions were eye pain (stinging), blurred vision, eye irritation (burning), eye redness, discomfort at instillation site (sticky eyes or sticky eyelids), eye itching, watery eyes, and medicine deposit on the eye lashes or around the eyes.

Please click [here](#) for full Prescribing Information and Instructions For Use.

What is CYSTADROPS (cysteamine ophthalmic solution) 0.37%?

CYSTADROPS is a viscous, or thick, cystine-depleting ophthalmic solution indicated for the treatment of corneal cystine crystal deposits in adults and children living with cystinosis. Cystinosis is a complex, rare disease requiring patients and caregivers to manage multiple different medications every day.

CYSTADROPS is the first and only FDA-approved cysteamine eye drop formulation applied four times a day during waking hours. CYSTADROPS can be stored at room temperature for up to seven days after opening.

Indications and Usage

CYSTADROPS (cysteamine ophthalmic solution) 0.37% is a cystine-depleting agent indicated for the treatment of corneal cystine crystal deposits in adults and children with cystinosis.

Important Safety Information

- To minimize the risk of contamination, do not touch the dropper tip to any surface. Keep bottle tightly closed when not in use.
- A condition where the pressure inside the skull increases for unknown reasons has been reported with cysteamine taken by mouth or cysteamine eye drops (used at the same time as cysteamine taken by mouth). This condition went away with the addition of medicine that increases the production of urine.

- Contains the preservative benzalkonium chloride. Contact with soft contact lenses should be avoided. Remove contact lenses prior to application. Lenses may be reinserted 15 minutes following administration.
- The most common side effects are eye pain (stinging), blurred vision, eye irritation (burning), eye redness, discomfort at instillation site (sticky eyes or sticky eyelids), eye itching, watery eyes, medicine deposit on the eye lashes or around the eyes.
- To report SUSPECTED SIDE EFFECTS, contact Recordati Rare Diseases Inc. at 1-888-575-8344, or FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

For more information, visit www.cystadrops.com.

About Cystinosis

Cystinosis is a rare genetic disorder affecting multiple organs and systems that most frequently begins in infancy. Cystinosis requires patients and caregivers to manage multiple different medications every day. It is estimated that approximately 600 people in the U.S. have cystinosis. Cystinosis is caused by a mutation in the CTNS gene that results in impaired transport of the amino acid cystine out of lysosomes in cells. This, in turn, leads to formation and accumulation of cystine crystals in cells, causing damage to organs throughout the body and significant impact on the eyes.

The cornea, or front layer of the eye, is the part of the eye that may be most affected. The first and most frequently reported ocular symptom is photophobia -- sensitivity to light that results in discomfort. It is thought that photophobia is mainly due to the presence of corneal cystine crystals that cause light entering the eye to scatter. As the disease progresses, ocular symptoms increase in number and intensity, daily activities become more difficult to carry out, and severe complications may develop, including visual impairment and potential corneal transplant.

About Recordati Rare Diseases Inc.

Recordati Rare Diseases Inc. is a biopharmaceutical company committed to providing often-overlooked orphan therapies to the underserved rare disease communities of the United States. Recordati Rare Diseases is part of the Recordati Group, a public international pharmaceutical company committed to the research and development of new specialties with a focus on treatments for rare diseases.

Recordati Rare Diseases' mission is to reduce the impact of extremely rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis and expand availability of treatments for people with rare diseases.

The company's U.S. corporate headquarters is located in Lebanon, NJ, with global headquarter offices located in Milan, Italy.

For a full list of products please click here: <http://www.recordatirarediseases.com/us/products>.

For additional information, please visit our website: www.recordatirarediseases.com/us.

Media Contact:

Elissa Johnsen

EJJ Communications, LLC for Recordati

312-285-3203

ellijohnsen@ejjcommunications.com

Statements contained in this release, other than historical facts, are "forward-looking statements" (as such term is defined in the Private Securities Litigation Reform Act of 1995). These statements are based on currently available information, on current best estimates, and on assumptions believed to be reasonable. This information, these estimates and assumptions may prove to be incomplete or erroneous, and involve numerous risks and uncertainties, beyond the Company's control. Hence, actual results may differ materially from those expressed or implied by such forward-looking statements. All mentions and descriptions of Recordati products are intended solely as information on the general nature of the company's activities and are not intended to indicate the advisability of administering any product in any particular instance.

November 12, 2020

Dear Cystinosis Community:

A few weeks ago, we informed you that CYSTARAN® (cysteamine ophthalmic solution) 0.44% eye drops would enter into shortage. We are very sorry for this disruption. Leadiant has a longstanding commitment to you, and we have always strived to be transparent and keep you informed on any news that may affect your treatment.

Yesterday, all the currently available supply of CYSTARAN in our inventory was delivered to patients. As of today, AllianceRx Walgreens Prime is unable to fulfill any orders of CYSTARAN, and while the shortage is temporary, at the moment we cannot provide an exact date of when CYSTARAN will be available again.

Everyone at Leadiant understands the importance of continuous treatment of the eye to treat the accumulation of corneal crystals in people with cystinosis. We are deeply sorry that we cannot meet your or your loved one's need for treatment.

While this shortage is temporary and many of the factors that contributed to it are beyond our control, we know you put your trust in Leadiant. It is our duty to resolve this issue as quickly as possible.

During the shortage, our commitment to you is to continue our work in restoring the supply of CYSTARAN as quickly as possible. Once we know when we can restore the supply of CYSTARAN, we will inform the entire community.

We will also provide you and the advocacy groups with regular updates on what we are doing to bring CYSTARAN back to patients quickly. We know you may have questions about this and want you to know that we are here. If you have questions about your treatment, call your physician. If you have questions regarding the shortage, please contact Lesli King at Lesli.King@leadiant.com or at 301-670-5450.

Please know that despite this setback, we continue our commitment to the cystinosis community. We have been your partner for more than 20 years and will remain with you for as long as the need exists.

Thank you,



Michael Minarich, Chief Executive Officer

Lesli King, Director, Marketing

Gianfranco Fornasini, Senior Vice President, Scientific Affairs

Giuseppe Izzi, Vice President, Administration and Chief Financial Officer

Giuseppe Testa, Vice President, Product Development

Joseph Wiley, MD, Vice President, Medical Affairs, Drug Safety and Pharmacovigilance

Welcome New Board Members



**Jonathan Dicks,
Vice President of
Development**

Jonathan is a critical care nurse, working his way towards

a Masters in the Family Nurse Practitioner track at the University of Cincinnati. This year Jonathan with three other founding members launched Revive Strength and Wellness, a client-focused personal training facility in Oakley. When not training clients or rotating in various outpatient settings Jonathan is a doting father and husband to his two little ones Elle and Finn, and “the bedrock of it all” his incredible wife Shirley. Married for eight years the family of three, expecting their second child were finally given a correct diagnosis for Eleanor, and thus began their “new normal.” Since that time Jonathan has been active in the CRN with his family making the trip to Philadelphia for their first CRN conference and representing his home state of Ohio with CRN at this year’s Rare Disease Week on Capitol Hill. Jonathan is an avid outdoorsman, reader, and fitness enthusiast who loves to cook, and enjoys spending as much time as he can with his family.

We all came to this place in one way or the other. For whatever reason or circumstance, cystinosis is the tie that binds us all. Our passion and drive to be a voice for those who have none calls us to do more, always more. I find myself here willing and able to lend all I can to this incredible community of link-minded souls. – Jonathan Dicks



**Andrea Carr,
Director**

Andrea Carr is originally from New Hampshire, but now lives in Dexter, NY

with her husband Jason and stepson Maddox. Jason and Andrea have two grandchildren, Lillian (“Bug”), 5 and Benson (“Moose”) 3. Andrea works at a public library as an Executive Assistant to the Director, but wears many hats.

Aside from her job at the library, Mrs. Carr spends a lot of time with family and friends, as well as working on their 57 mile relay called “Miles for Moose” to raise funds for cystinosis, because our grandson was diagnosed in 2018.



**Melanie Vachon,
Director**

Melanie Vachon is a dental assistant from Derby, Kansas. She is a mother of two

boys, Zander-9, and Logan-almost 8. Logan was diagnosed with cystinosis just before his first birthday. The diagnosis was hard, life changing; it was very tough for a while. The struggle was real, but Logan is now not only stable, but thriving!

Melanie has had the pleasure of attending several CRN/cystinosis events in the past few years, including the CRN conference in Utah, and smaller cystinosis meet-ups in LA, St. Louis and Wichita. She was also given the opportunity to represent CRN at Rare Disease Week on Capitol Hill this past February. The experience at Rare Disease Week, representing and advocating for our cystinosis community on The Hill, lit a fire inside of her.



Tim Wyman, Director

Timothy Wyman, CFP®, JD, is the Managing Partner and CERTIFIED FINANCIAL

PLANNER™ professional at Center for Financial Planning, Inc.® Tim earned a place on Forbes’ Best-In-State Wealth Advisors List in Michigan in 2019 for the second consecutive year. He was also named a 2019 Financial Times 400 Top Financial Advisor.

A leader in his profession, Tim has served on the National Board of Directors for the 28,000-member Financial Planning Association. He frequently speaks to organizations and businesses on financial planning topics. A frequent contributor to national and local media, he has made appearances on Good Morning America Weekend Edition and WDIV Channel 4 News, and has contributed to articles in Forbes, The Wall Street Journal, Money Magazine, SmartMoney, The Chicago Tribune, and other national publications.

Tim has been an active community volunteer over his 25+ year career serving several organizations. He currently serves on the Albion College Endowment Investment Committee.

Tim and his wife, Jen, have three children. The oldest, Matt, graduated from the University of Kansas where he was a placekicker for Jayhawks football team. The middle child, Jack, a U.S. Army 2nd Lieutenant, lives with his wife Libby in Washington. He also graduated from Tim and Jen’s alma mater, Albion College, and captained the college’s baseball team. The youngest child, Kacy, is a student at Bloomfield Hills High School and looks forward to her college years in the near future.

Development Update

By Jonathan Dicks, Vice President of Development

Two years in. Two years almost to the exact day when our burgeoning family's lives would change forever. In the acute phase of Eleanor's diagnosis I couldn't see anything but darkness. The silver lining wasn't there, and if there was a light at the end of the tunnel, our collective grief blocked those rays of hope. All we could think about was what to do next? Shirley was six months pregnant with our son, Finn, and we realized the nature of this disease and its genetic inheritance immediately. Needless to say we walked on eggshells for the next three months in anticipation, with a resolve to tackle whatever news we were given. Cystinosis wasn't thinking for one minute about taking a day off, we weren't either.

Two years has given so much perspective allowing us to realize the many blessings we have to be thankful for. It has also challenged us to realize we cannot live this life without the help of "our village." I didn't truly understand that until Elle was properly diagnosed and we truly understood what the future could possibly bring with it. We were managing the brand new expectations of our lives. In totality the path seemed insurmountable, but in little chunks we were seeing positive movements. Some days the highs and lows changed drastically, but we soon began to remove the really bad days with better days. When Shirley and I officially joined the CRN and attended our first family conference we knew we had found a home.

All I could think about was how do I give to a community that gave us so much in our deepest time of need?



How can I best support those battling this disease currently, and honor those beautiful souls represented in the twinkling white stars that surround our green ribbon? I immediately began to search for ways I could be at the center of my daughter's care and still make meaningful moves for not only her, but for all the incredible warriors and their families with this diagnosis. I knew there was space for me somewhere, but I was not certain where I would fit so I chose to say "yes" to anything that came my way.

When Clinton gave me a call to assess my interest in representing CRN and my home state of Ohio at the 2019 Rare Disease Week on Capitol Hill I immediately accepted. When he then floated the idea of



Jonathan Dicks (right) at the 2019 Rare Disease Day in Washington, DC

working in a more focused fashion on the executive board, I did the same. I didn't know what or where he felt I would have the best fit and greatest impact, I was just excited to be given the opportunity to serve a cause that had become our life's passion. He and I both knew I was onboard for life.

Two years in and now in the role of Vice President of Development I have found that home. The role has been a ridiculously busy undertaking from the very first conference call, but that's quite an amazing truth to realize. It means we are seeing a litany of energized families in our small community stepping into advocacy roles and fundraising events. With the help of some incredible behind-the-scenes work from truly amazing people this development role has blossomed into an interconnected role of helping families interested in fundraising to do so in a stream-lined and stepped fashion that takes the fear out of the question "what can I do?"



4TH ANNUAL

Cystinosis Awareness Day

MAY 7, 2021

Update: Adult Leadership Advisory Board

By Cheryl Simoens, ALAB Chairperson



I cannot believe how quickly time passes and that winter is upon us already. As I reflect over the past year as

Chairperson with ALAB, I recognize the dedication, collaboration, and growth this group has demonstrated in providing support, education, and mentorship to the entire cystinosis community. We have learned, shared laughter and built friendships in each other that will not be forgotten. I am very proud of the work we put in to develop and brand ALAB and create project initiatives from fruition to execution. With the launch of our three projects; Cystinosis Rare: A



ADULT LEADERSHIP ADVISORY BOARD

Journey into the Unknown Podcast, Cystinosis Sessions, a virtual face-to-face platform for discussions and CystinosisTEENS, a private Instagram account, there is something for everyone to participate in, listen to

and feel more connected to the entire cystinosis community across the world.

Below are further details and updates on the launches and content of these projects.

Cystinosis Rare: A Journey into the Unknown Podcast



Cystinosis Rare Podcast released the third episode in two parts in August 2020 focusing on transitioning from youth to adulthood. Part 1 included a panel of young adults and adults from the cystinosis community speaking to their own experiences on transitioning, struggles and challenges, coping strategies and how they addressed their own needs and moved to independence and resilience.

Part 2 included a panel of medical professionals; Dr. Paul Grimm, MD with the Lucile Packard Children's Hospital at Stanford University and Dori Rivera, PAM (Patient Access Manager) with Horizon Therapeutics, discussing tips on how to manage medical, social, and educational needs, guidance in the medical system, including protection during COVID, and tools, resources and strategies for smooth transitioning into adulthood. The fourth and fifth episodes were released in November 2020 with Maya Doyle, MSW, Ph.D., LCSW with Quinnipiac University, moderating both episodes focusing

on bullying, social implications, negative biases, and teasing. Episode 4 consisted of a panel of teens and young adults/adults sharing their stories on personal situations and experiences with these issues, how it shaped them and coping tools and strategies they learned and utilized. The fifth episode welcomed a panel of parents with children who have cystinosis discussing how they managed and continue to manage their child's needs, their own experiences with bullying or social implications and hopes for their children in the future.

CystinosisTEENS Instagram Account

The Instagram account was created just for teens and young adults. It is doing well with ongoing, regular spotlight posts highlighting teens and young adults' successes, interests, and achievements, allowing followers to be reminded that they are not their illness and the potential is



endless! The account also provides promotional posts focusing on ALABs project initiative updates and reminders, fundraising efforts from both ALAB and CRN, and

opportunities for the entire cystinosis community including scholarship funding, and inspirational quotes.

Cystinosis Sessions

ALAB members have been busy working on monthly virtual Zoom sessions focusing on connecting cystinosis community members from all over the world to join in conversation. Throughout the months, topics have included mental health; PTSD, relatable, shared stories and coping practices, including therapy; exercise and keeping active, which had the conversation focusing on types of exercise, areas in the body that muscle

deterioration has been noticed, and a personal trainer as special guest to answer questions; and nutrition and diet among the community. Cystinosis Sessions has seen participants from around the world including Germany, Mexico, Kuwait, Canada, and the US. If you are a young adult or adult with cystinosis interested in getting more involved and joining the conversation, this is a great place to start!

Contact Cheryl at cheryl.simoens@gmail.com.



Exciting Engagement Opportunities



In July 2020, ALAB members had the opportunity to participate in the 6th Annual Dublin Cystinosis Workshop as keynote speakers in a research symposium held by Cystinosis Ireland. The presentation focused on development and programming, needs of adults with cystinosis, and the importance of listening to

the patient voice. The keynote was delivered to an esteemed group of physician researchers, clinicians, and organization leaders from around the world. ALAB members advocated for adults living with cystinosis through sharing their mission, goals and needs of the community as we enter a new world where the adults are becoming the voice and the experts that drive momentum, success and planning.

It is with great excitement that I announce ALAB had their first Open Call for 2021 Applications

for new members in October 2020 with announcements being made by early 2021. It is an honor to be working alongside ALAB and CRN in collaboration of the intake process from selection to onboarding and training of new members. I look forward to learning from this new group, mentoring new faces and collaborating with a fresh set of ideas, perspectives and strategies. The year ahead will be bright and full of excitement, growth and transformation.

Walk in My Shoes – Documentary

By Clinton Moore, President

At first the thought of creating a documentary about a walk event that I do each year seemed really simple. And it probably would have been. As the discussions began and then the planning I soon realized that what I truly wanted to share was so much more than a walk and that the walk was only a very small part of it.

I wanted to share MY story, my SON'S story, and my WIFE'S story. Everyone's story and perspective about a rare disease is different. A rare disease that affects only a couple thousand people affects each of them differently. In every single person you will find differences even though it's the same disease.

The filming began with me just holding a cell phone a recording myself. Just to have small short clips to add in here and there. The film crew showed up and started filming on the day before the walk. Hours and hours and hours of footage were gathered. Some material was interviews with other walkers and some just shots of everyone walking around the track. All day and all night, the cameras rolled until we completed the 57 miles. The next morning, assisted by crutches, I worked my way into the makeshift studio for another full day of on camera interviews. Once we ended I thought we were finished filming. I could not have been more wrong.

After reviewing all gathered footage, Richard Hemmingway (producer) called me and told me what we could create with what we had. I knew right away that we needed more. Much more. He then gathered up and found his way to Philadelphia for the CRN family conference in July (2019) with big plans to finish shooting. After an

unfortunate series of events we had to postpone. My son's health was failing and we now had to turn our attention to him and his doctors to figure out what was going on. The documentary had to be put on hold as we could not get the footage we wanted to make this a great film. Nor did we have the time.

After a few months of vigorous testing, doctor visits, second opinions and surgeries, things started to look up. Now we were ready to finish this thing up. Round three was scheduled and soon I found my home once again filled with cameras and lighting equipment. Three days later cameras were packed up and filming was complete.

Then came editing. If we used every minute of footage we had then the film would be close to 200 hours long. Next, I was introduced to the grueling task of removing footage and creating a finish product. This takes months. Then when you think you're done you watch it and make more changes. Then you decide to add something and then remove something. I honestly think I could have worked on this project forever. But at some point you have to stop and love what you created.

Creating the "Walk In My Shoes" documentary was challenging. It was emotionally challenging as well as

mentally. I am completely satisfied with the final product. It shows OUR story. Just the way I wanted it to. It shows our "highs" and our "lows." It shows how WE deal with cystinosis. Blended in certain spots you meet other people to get THEIR perspective. I am extremely proud of this documentary and proud of all the people who were a part of it.

Thank you to everyone that took the time to watch and to everyone for the overwhelming and incredible amount of support and love that came right after its release.

To view the full-length documentary, please visit the Cystinosis Research Network channel on [youtube.com](https://www.youtube.com).



Clinton Moore on set during the "Walk In My Shoes" Documentary Satellite Media Tour

Facebook Fundraising: Recognizing Your Contributions

Online giving has grown consistently year over year. The cystinosis community has embraced this option, especially when it comes to Facebook fundraisers. If you are among the platform's 2.6 billion monthly active users, you've probably been encouraged to create a birthday fundraiser for your favorite charity or you've been prompted to share a campaign to fund emergency relief.

While Facebook fundraising has expanded our donor reach, recognizing those donors has been unsuccessful. Ideally, we would fill the annual donor honor roll with the name of each and every person who has selflessly contributed. Due to Facebook privacy policies, donor information remains anonymous. In 2019, we received almost \$35,000* from Facebook fundraisers. These often small acts of kindness add up, creating funding for research and support programs.

Thank you for your continued support!

*Facebook contributions are received via Network for Good.



Facebook fundraisers have become a good source of fundraising thanks to the ease of use. If you have a Facebook account, the platform may prompt you to begin a birthday fundraiser. In November 2017, all fees were abolished and now 100% of donations go towards your selected nonprofit. Here's how to get started:

1. Click  **Fundraisers** in the left menu of your News Feed.
2. Click **+Raise Money**.
3. Under **Nonprofit** select **Cystinosis Research Network**.
4. Choose a cover photo and fill in the fundraiser details.
5. Click **Create**.

CRN and eBay for Charity



eBay for Charity has partnered with the PayPal Giving Fund to make it easy for sellers to donate 10% to 100% of your item's final sale price to a certified charity.

1. SELECT CHARITY

Seller picks the charity and the donation percentage when listing an item.

2. SELL & SHIP

Item sells. Buyer pays full amount to seller. Seller ships item to the buyer.

3. DONATE

After the transaction is complete (approx 21 days), PayPal Giving Fund

will automatically collect the donation from the seller's PayPal account* Once a month PayPal Giving Fund will combine and deliver 100% of all donations collected for that charity.

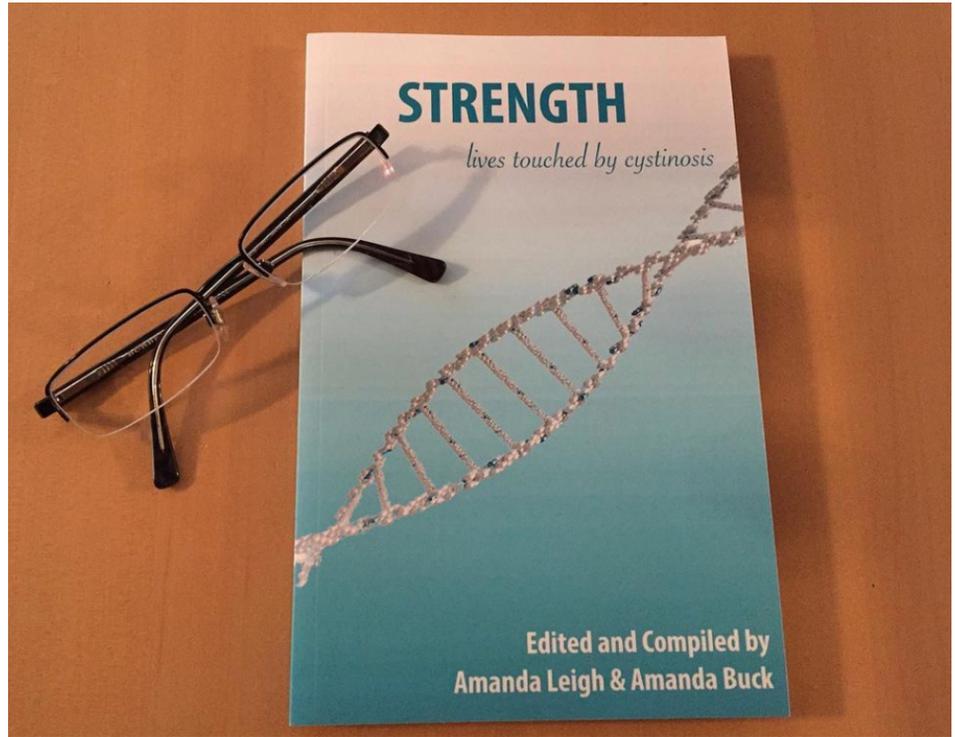
*If your donation cannot be collected automatically then you will be emailed an invoice from PayPal Giving Fund requesting payment for the donation.

CRN is a certified charity within this program. You can learn more at <https://ebay.to/2YkSxQ9>.

Strength: Lives Touched by Cystinosis

Each Cystinosis journey is different. However, this collection of stories reminds us of a trait many have in common: Strength. Hear from over 20 individuals and loved ones impacted by cystinosis. Amanda Buck (cystinosis caregiver) and Amanda Leigh (adult living with cystinosis) deliver this labor of love on behalf of the cystinosis community, but it is intended for EVERYONE who has ever experienced the great pains and joys of life.

Please consider purchasing a copy today. All proceeds benefit the Cystinosis Research Network and move us one step closer to finding a cure. Available through [amazon.com](https://www.amazon.com).



Take the “Know Your Cystinosis” Quiz

When were you diagnosed with cystinosis? _____

Have you/when did you receive dialysis and/or a transplant? _____

What are your medications? _____

What is each medication for? _____

When do you take them? _____

Any side effects? _____

Are you allergic to anything? _____

Any major surgeries or hospitalizations? _____

Who are your doctors and how do you reach them? _____

Knowing the answers to these questions is important as you prepare to transition to a new medical team. They are also vital to know if you are involved in after-school activities, out with friends or traveling. In an emergency situation, like a sudden illness episode, a sports injury, or a car accident, you must be able to tell medical personnel about your health and your medications. You can find additional tools at [cystinosis.org](https://www.cystinosis.org) under Support & Resources, Education section.

Cystinosis Research Network
P.O. Box 702
Lake Forest, IL 60045-9998

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P.O. Box 702
Lake Forest, IL 60045-9998
www.cystinosis.org

Phone 847-735-0471

Fax 847-235-2773

Email info@cystinosis.org

Editor Clair Johnstone

Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cystine, to accumulate in various organs of the body, including the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine. The availability of cysteamine medical therapy has dramatically improved the natural history of cystinosis so that well treated cystinosis patients can live into adulthood.

CRN VISION

The Cystinosis Research Network's vision is the acceleration of the discovery of a cure, development of improved treatments, and enhancement of quality of life for those with cystinosis.

CRN MISSION

The Cystinosis Research Network is a volunteer, non-profit organization dedicated to advocating and providing financial support for research, providing family assistance and educating the public and medical communities about cystinosis.