CYSTINOSIS RESEARCH NETWORK

Volume 13, Issue 1
Spring/Summer 2020

THE CYSTINOSIS Advocate

Inside This Issue

Visit our YouTube channel at bit.ly/2XG8fbk

The 2021 CRN Family Conference will be hosted in Nashville, Tennessee!

This charming location features unique music, culture, food, landmarks, and southern hospitality. We hope you can join us as we bring together cystinosis individuals and families from around the world. Take a virtual tour of the **Nashville Marriott** where all conference activities will take place at https://bit.ly/2Ae9x2H

Mark your calendars for July 15 – 19, 2021!

Please check back for an agenda and additional updates later this year. We look forward to seeing you in Summer 2021.

The President's Letter

Clinton and Chandler Moore.

I think it's very safe to say that the world is a much different place today than it was when I wrote the last president's letter. COVID-19 has swept every social media platform, news coverage, and very apparent when you go into public seeing everyone wearing masks and social distancing. The overall feel of the world is just very different and challenging in many new ways than it was before. Or at least that is what you hear from so many people. I have to say, I agree, and disagree. Let me explain...

For many people, this is a scary time. Fear of contracting the virus overwhelms a huge percent of the world's population. Having to social distance is a burden and inconvenience. Having to find new ways of doing the things you like to do is frustrating. These are the things that need to be done. Following the recommended guidelines is what will bring us out of this the fastest.

From a rare disease patient, caregiver, or family member... not much has changed. Millions of rare disease patients including cystinosis patients are immunocompromised which has forced them to transform their lifestyles long before COVID-19 told us we had to. I've heard many cystinosis families say "welcome to our world".

From a nonprofit standpoint many things have remained the same. We've always done a majority of our planning and meetings through conference calls. Our overall focus and goals have remained the same and being this is our off conference year, we haven't had to cancel or reorganize the structure of that. Our regional in-person meetings are being reimagined in a safe, virtual way to continue connecting families despite the COVID-19 restrictions (see page 26 for details).

We have been monitoring the situation very closely and have been getting updates from many of our key opinion leaders to stay ahead of any specific COVID related needs the cystinosis community may have. This will continue throughout the course of the pandemic regardless of its length.

In the meantime the Cystinosis Research Network continues to be your advocacy group just as we were before COVID. Just as we will continue to be after this pandemic is a thing of the past. Reach out to myself or any member for anything COVID related or non-COVID related. We are here to help in any way we can.

Stay safe, stay healthy, and remember, even if alone... we will get through this together.

Clinton Moore

Bill Gahl receives HHS award

By Prabarna Ganguly, Ph.D., National Human Genome Research Institute

Dr. Gahl honored for his leading efforts to diagnose and treat individuals with rare and undiagnosed diseases.

The Department of Health and Human Services (HHS) has awarded National Human Genome Research Institute (NHGRI) senior investigator Bill Gahl, M.D., Ph.D., the prestigious Career Achievement Award. This award recognizes his undeterred commitment to providing answers and possible treatments for people suffering from unknown and rare genetic conditions.

One of the highest honors bestowed by HHS, the Career Achievement Award "recognizes HHS employees with 10 or more years of service at HHS for dedication to the Department with a record of outstanding achievements with a high standard of excellence and dedication throughout their HHS career." Dr. Gahl is one of five recipients of the HHS Career Achievement Award this year.

"Bill's many years of contributions to genetics and genomics research, coupled with his passionate and dedicated care of numerous patients with rare diseases, make him overwhelmingly worthy of this career achievement award," said NHGRI Director Eric Green, M.D., Ph.D. "He exemplifies the best of a dedicated physician-scientist and serves as a role model for many others. NHGRI has been truly fortunate to have him within our ranks."

Dr. Gahl's signature achievement was the founding of the **National Institutes of Health (NIH) Undiagnosed Diseases Program** (UDP) in 2008. The aim of the UDP is both simple yet extremely difficult - finding answers to the most puzzling medical cases from around the world. Dr. Gahl has brought to the program some of the best scientific minds in medicine and genomics. In 2012, NIH expanded the program to form a nationwide **Undiagnosed Diseases Network** (UDN), which includes 12 clinical sites and supporting facilities across the country. The UDP's first discovery involved uncovering the genetic basis of the rare disease Arterial Calcification due to Deficiency of CD73 (or ACDC, as the team called it). UDP published their work in the New England Journal of Medicine in 2011. To date, they have discovered 23 new genetic disorders and disease phenotypes.

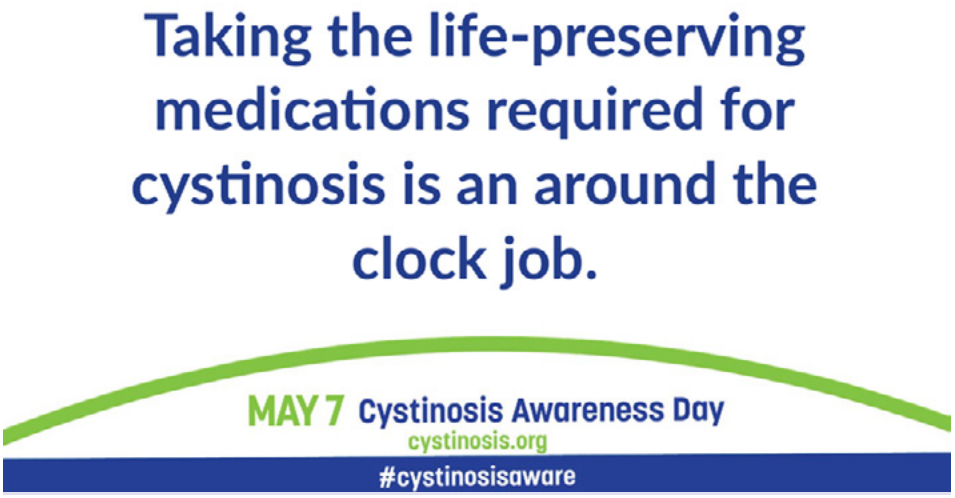
Bill Gahl, M.D, Ph.D., Senior Investigator, NHGRI. Credit: Maggie Bartlett, NHGRI.

"The UDP has been an extraordinary engine of discovery that has markedly accelerated biomedical research while providing answers for our patients," remarked Dr. Dan Kastner, scientific director of the NHGRI Intramural Research Program.

For 16 years, Dr. Gahl served as NHGRI's Clinical Director, stepping down from that position in 2019. Colleagues and patient communities have often touted him to be a world-class medical geneticist, a premier researcher, a compassionate physician, and a visionary scientific leader.

"The HHS Career Achievement Award speaks to the incredible opportunities offered by the NIH Intramural Research Program, the Clinical Center, and the expert scientists and physicians who have been my colleagues and friends for more than a generation," said Dr. Gahl, responding to receiving the award. "To me, the NIH reflects the best that our government has to offer, and I am incredibly honored and grateful to be part of that."

3rd Annual Cystinosis Awareness Day
May 7, 2020



Cystinosis Warrior.

It is a title bestowed upon each and every individual who has lived with and continues to live with this disease. Different from many other titles, this one is not chosen, it is assigned at birth. It is not a badge you can strip away or denounce. The bodies of our cystinosis warriors fight a battle each and every minute. The Cystinosis Research Network is proud to honor you on Cystinosis Awareness Day, and every day as we march together to defeat this relentless disease.

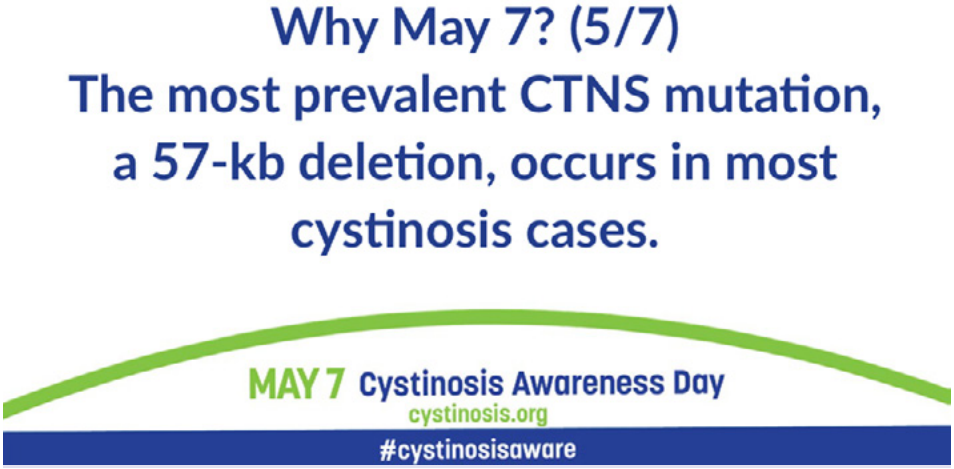
This year was the third anniversary of Cystinosis Awareness Day and it was nothing like the first two events. Our goals did not change: Educate as many as possible about cystinosis and raise funds for vital research. But the global landscape changed dramatically in the months leading up to awareness day. Never before has our generation encountered a pandemic like COVID-19. For our immunosuppressed warriors, the novel virus posed yet another threat. Although COVID-19 was new, the circumstances feel somewhat familiar.

Once again, we find ourselves in the middle of a battle we did not sign up for; one that will test our vigilance and determination.

So we moved onward focused on our targets – raise awareness and funding. Our multi-channel approach through email, the cystinosis.org website, and online platforms like Twitter, Instagram, Facebook, YouTube, and most recently, TikTok, were embraced beyond the cystinosis community. May 7th was recognized by partner groups including national and international disease organizations around the world. Seeing countries outside the U.S. like Ireland, Iran, Turkey, and Africa commemorating the day fills our hearts with an immense amount of pride. Through these efforts we reached over 50,000 people!

Leading up to the month of May we witnessed other non-profits being hit hard as donations dropped dramatically. During these uncertain times, any cause that didn't directly benefit COVID-19 relief seemed to take a back seat. Once again, our cystinosis warriors rose to the challenge. The 2020 awareness day campaign surpassed our fundraising goals. Early calculations estimate over $36,000 raised.

All of these achievements are possible because of you - our cystinosis family. From a young warrior demonstrating a g-tube feed to our adults providing hope though stories of "Succeeding with Cystinosis" [see pg 16], your ongoing support is important now more than ever. Thank you.



Cystinosis Awareness Day Advocacy

As “New” Cystinosis parents, a lot of the time we feel like we are on such a roller coaster with everything we do. We are learning everyday what this disease can do. The toll it can take. On us as parents, on a marriage and on a brave, strong little soul. I feel like we have really jumped into this world, this community of amazing, strong, supportive people, and we have been received by them. This alone has given us such gratitude and fulfillment.

Recently we were asked by the team at Avrobio to talk with their staff on what Cystinosis is really like. We would be talking to the team that focuses on gene therapy, and not just the Cystinosis team, but all those that work towards finding therapies and possible cures for a variety of rare diseases. Can you even imagine? A time of not worrying about levels, and liquid amounts, and getting up in the night, and even, maybe just for a little while, not having to hear the word Cystinosis!

When asked we were so excited. We love telling our story, and putting as much education abut Cystinosis out there! We were truly honored that they would want to hear all about our day to day life! It was incredibly meaningful to be able to give a glimpse about what life is really like for us on a regular basis. Both Brian and I work in fields that focus on others. Sometimes you need to see or hear first hand really what it is you’re working so hard for, to continue fighting. That was our goal in meeting with the Avrobio team, to give them that snapshot and help provide encouragement to continue their fight in finding a therapy that works. As a family we want to really show how their work, can effect families like ours, not just for a little bit, or for one point in time, but long term. To show their work matters and can make a difference and that continuing forward can change lives. We thank Lisa, Fernanda, and the team, so much for making that lunch and learn happen! We hope that more are to come and that we can continue learning and growing from one another! Thanks Avrobio for all you do for our Cystinosis family!

To the Meschke Family, From AVROBIO

Dear Chelsea and Brian,

We can’t thank you enough for taking time on Cystinosis Awareness Day to share your family’s story with all of us at AVROBIO. You taught us about Jaxon and what it’s like to be parents of a child with cystinosis. The daily challenges of mealtimes and managing so many medications, but also the joys of an energetic little boy who loves to play outside and jam out to ‘80s hard rock! It’s heart-warming to hear that Jaxon’s diagnosis does not define who he is, and we’re excited for the possibility that we just met a future rock star!

Your honesty, resilience, and love for your son touched and inspired every member of our organization. On behalf of AVROBIO, we would like to express our sincere gratitude for giving us this invaluable opportunity to educate and motivate our team as they work hard to find new ways to help families affected by cystinosis.

Meschke Family Christmas 2019

Brighter DAYS

Brought to you by the CYSTINOSIS RESEARCH NETWORK

Brighter Days is a brand new program available to our cystinosis community.

Go to our website (cystinosis.org) and look for the Brighter Days logo for details.

Virtual International Cystinosis Family Conference 2020 – the biggest ever cystinosis event!

Cystinosis Network Europe and Cystinosis Ireland hosted the first ever Virtual International Cystinosis Conference 2020 on 25 April 2020. Within the space of just 4 weeks, Cystinosis Network Europe and Cystinosis Ireland turned the disappointment of canceling the highly anticipated biennial International Cystinosis Conference 2020 (which had been scheduled to be held in Ireland in July in Dublin) and converted it into a hugely successful virtual event.

The virtual conference attracted an audience of more than 600 participants from 49 different countries in 20 time zones across the world. It was simultaneously translated into 7 different languages and featured presentations on a wide variety of clinical and research aspects of cystinosis from 16 world renowned clinicians, researchers and other healthcare professionals from USA, Canada, Ireland, Belgium, Germany, France, England and Scotland. Many of the conference logins included several members of families. We were joined from countries as far away as Australia, Mexico, Turkey, Egypt, USA and Canada. Many who joined would not have been able to travel to the original in-person conference that had been planned. One family in Australia listened in to the whole conference until 3 in the morning! Some participants had never before had an opportunity to listen to world class cystinosis experts speak and answer questions about their ultra-rare condition.

The keynote speaker was Dr Stephanie Cherqui, Associate Professor at the Department of Paediatrics, Division of Genetics of the University of California, San Diego, USA. Dr Cherqui's highly anticipated contribution to the conference focused on her pioneering research developing an autologous stem cell and gene therapy for people living with cystinosis. Last year, Dr Cherqui received FDA approval for a human Phase 1/2 clinical trial for this ground-breaking therapy and the first patient was transplanted last October. Dr Cherqui provided an update on the positive progress of this first patient who received modified version of their own stem cells which had been genetically engineered to produce and deliver functional cystinosin throughout the body. At three months post-transplant, the patient has shown no unexpected safety events and a very promising reduction in their

VIRTUAL
International Conference 2020
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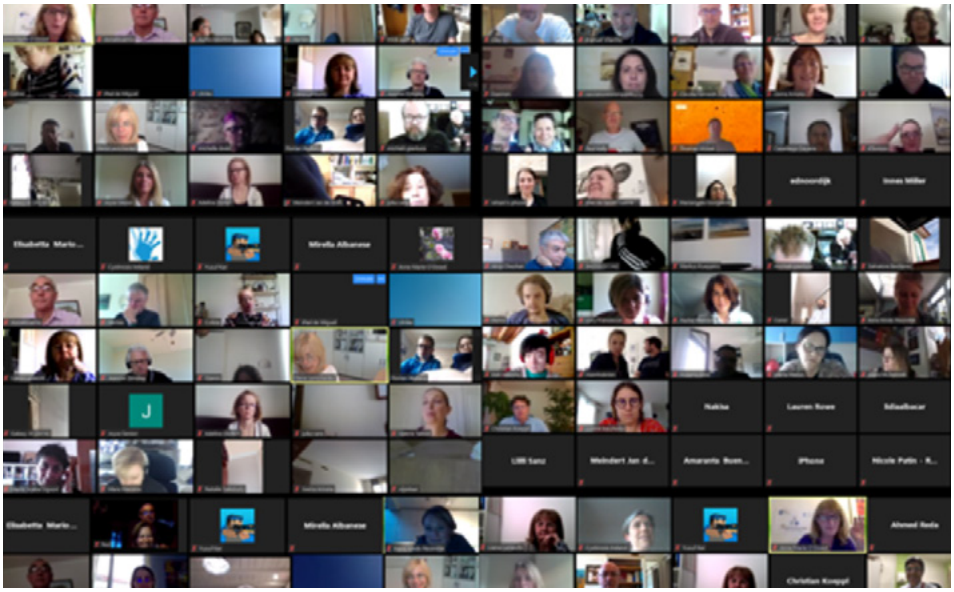


Some of the more than 600 virtual conference participants.

treatment burden. While recruitment onto the trial has been curtailed by the current Covid19 pandemic, it is hoped to enroll further participants in the future once circumstances permit.

The Virtual International Cystinosis Conference 2020 attracted a diverse mix of attendees including adults and families with many years' experience of living with cystinosis as well as families that have newly diagnosed young children and are just beginning their cystinosis journey.

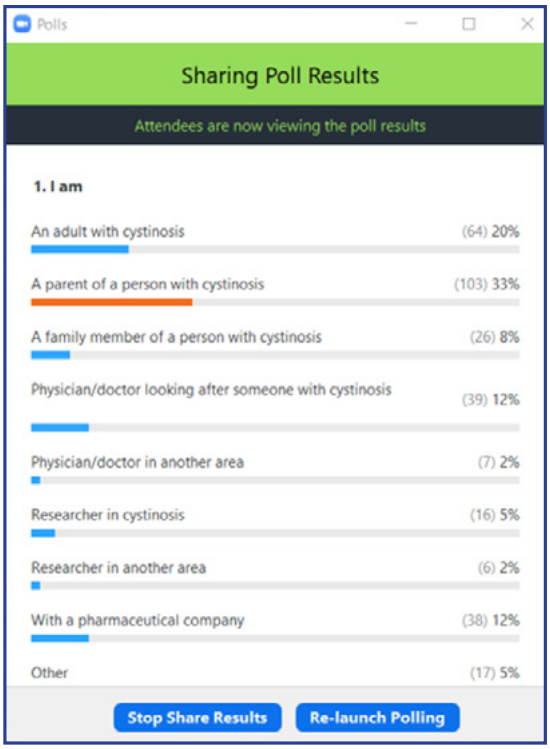
In order to address the needs of this diverse audience, the conference drew on a wide variety of expert contributors who focused on providing essential information about cystinosis as well as highlighting new knowledge and breakthroughs in our understanding and treatment of the disease. Speakers included:

Professor Elena Levtchenko (Conference Scientific Chairperson; Pediatric Nephrologist; University Hospitals Leuven, Belgium) who gave an overview of cystinosis and how to manage it; Professor Don Cairns (Head of School of Pharmacy and Life Sciences, Robert Gordon University, Scotland) who explained to participants how the current cystinosis drugs Cystagon and Procysbi work and why they are important as well as highlighting potential new drug therapies that are currently being developed; Dr Joyce Senior (Department of Education, University College Dublin) discussed the educational needs of children living with serious chronic illnesses and the importance of working with schools to help them understand these needs.

A number of the conference sessions focused on the complications that arise with cystinosis. Dr Rachel Bishop (Clinical Ophthalmologist at the National Eye Institute, National Institutes of Health, Maryland USA) spoke about how to effectively manage complications of the eye in cystinosis. Professor Anuj Chauhan (Head of Department of Chemical and Biological Engineering, Colorado School of Mines, USA) presented on his exciting and novel research which aims to improve treatment of ocular manifestations of cystinosis by using contact lenses to deliver the cysteamine to the eye.

Professor Justine Bacchetta, Professor of Paediatrics at the CHU de Lyon – Hôpital Femme-Mère Enfant in Lyon, France and Mr Christian Köppl, Physiotherapist at the Centre of Social Paediatrics in Traunstein, Germany presented on bone and muscle complications associated with cystinosis focusing on the underlying mechanisms why these occur and the importance of exercise to help reduce the effects of the disease. Renowned pediatric nephrologist Dr Paul Grimm, (Stanford University School of Medicine, USA) gave an important presentation on explaining how and why cystinosis affects the kidneys, and how patients can maintain their kidney health to the greatest extent possible prior to or post transplantation. Dr Aude Servais, Senior Nephrologist at the Department of Adult Nephrology and Transplantation, Necker Hospital, at Paris Descartes University, in France and Dr Ahmed Reda, Postdoctoral Fellow, at the University Hospitals in Leuven Belgium both spoke about fertility in female and male patients living with cystinosis respectively.

In addition to clinically-oriented presentations, the conference also spotlighted exciting new developments in cystinosis research. Apart from Dr Stephanie Cherqui's keynote presentation, other research highlights included: an overview of cystinosis research over the years and current research on the use of novel drug combinations to treat cystinosis which was delivered by Dr Patrick Harrison, University College Cork; Professor Emeritus Herbie Newell of the University of Sunderland, who presented on the development of CF-10 an inactive form of cysteamine which is activated at the cell surface and which should lead to smaller and less frequent doses, with fewer side effects; Professor Paul Goodyer of McGill University, Montreal Canada who presented on another exciting



new drug therapy being developed by his research team which uses aminoglycosides to treat patients who have a specific cystinosis nonsense mutation; and a presentation by Professor Minnie Sarwal, Professor in Residence of Surgery, Medicine and Paediatrics at the University of California, San Francisco on a novel urine test which is being developed by her research team to track kidney injury and transplant rejection in patients with cystinosis.

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

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

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

Cystinosis Network Europe thanks all those involved in delivering this conference including all of the speakers, clinical experts and researchers who contributed to the conference, the organizers Cystinosis Ireland, the sponsors of the event and the scientific Chairperson, Professor Elena Levtchenko.

A recording of the conference will be available soon on www.cystinosis.ie

Worldwide Cystinosis Community Advisory Board

In other work, Cystinosis Network Europe, supported by Eurordis, is continuing to work with researchers and healthcare companies through the Worldwide Cystinosis Community Advisory Board (CAB). The CAB is led by patient experts – people living with cystinosis or their family members – who are trained and knowledgeable in the field of research and clinical trials.

The CAB enables the member organizations to interact in a structured and transparent way with treatment and medication researchers and developers to ensure patients are consulted as patient investigators in the development of treatments and therapies that will have an impact on their lives. The researchers and developers obtain valuable information and insights on areas including the efficacy of their informed consent process to the implementation of proposed research protocols, which they would not otherwise get about the community demographic they are targeting. For more information on the CAB and any of the work of CNE, please contact denise.dunne@cystinosis-europe.eu.

COVID-19 and the Cystinosis Community

In March of this year the COVID-19 pandemic changed life for everyone around the world. It quickly became clear that CRN as an advocacy organization needed to provide up to date information to our community, in particular given our specific concerns surrounding immunosuppression and chronic kidney disease.

CRN has provided a variety of resources to help navigate this pandemic, including the Adult Leadership Advisory Board's (ALAB) most recent edition of the podcast, Cystinosis Rare: A Journey Into the Unknown, which can be accessed on CRN's YouTube channel at http://bit.ly/2XG8fbk. In addition, ALAB hosted their first video meetup in the Cystinosis Sessions series, which featured CRN board member and nurse Anna Pruitt providing up to date information on the virus and guiding discussion on the topic.

CRN's advisory board members wanted to capture information on how the virus was affecting those with cystinosis. To that end, a survey was created and distributed in April to learn more about COVID-19 and its impact within the cystinosis community. Our goal was to gather your input during this time to provide valuable insights for healthcare professionals and the general cystinosis population. When the survey closed in mid-April, a total of 107 responses had been obtained, 69 from US, the rest international. At that point, of the caregivers and individuals with cystinosis who completed the survey, 3 had been tested for COVID-19: 2 individuals had tested negative and 1 tested positive for COVID-19. At that point, no one had been hospitalized as a result of the virus.

Of note, 95% of respondents reported that they were either "highly worried" or "somewhat worried" about the health risks of those with cystinosis and COVID-19. 75% reported being either highly or somewhat worried about the financial impact (loss of job, insurance, etc.) of the virus.

A follow up survey will be issued in the near future to assess any changes in prevalence of the virus in our community and to gather more insights into how the pandemic is affecting the lives of our families and individuals with cystinosis.

The National Organization for Rare Disorders (NORD) has launched a COVID-19 Relief Program. For more information on available assistance please go to https://rarediseases.org/COVID-19/.

In addition, NORD has an ongoing Patient Assistance program specifically for those living with cystinosis. More information can be found at http://bit.ly/2Xi6T1A.

For more information and other support resources please contact Jen Wyman, VP Family Support at jwyman@cystinosis.org.

COVID-19 Perspective

By Tory Kruse

Since COVID-19 has hit, so often the media is using the phrase “our new normal”. Raising a child with cystinosis, we’ve adjusted to rounds of medications, eye drops, tube feedings, and many doctor visits as our family’s “normal”. My mom’s a teacher and my two younger brothers are teachers. But while my husband’s long work hours has made it most beneficial for me to be a stay at home mom for the past four years, I did not EVER plan (nor have ANY desire) on being a teacher OR homeschooling a fourth grader and first grader while caring for an eight month old baby!

Avery, Braxton and Carter Kruse.

Our daughter, Avery, is a fourth grader, and is pretty independent. She accomplishes her school work each morning with little difficulty. Well, other than, “Mom, the internet’s being slow!” (I know, everyone’s is, honey) “Mom, the baby is in my room!” (Shut your door, honey). “Mom...!” Carter, our first grader, with cystinosis, faces more challenges. Carter has developmental delays, ADD and autism. Carter’s “normal” includes attending a school for students with developmental disabilities and he’s on an IEP, which not only encompasses academic goals, but also intense Speech Therapy, Occupational Therapy, and Physical Therapy. Each day, the kids’ teachers are amazing and provide them with wonderful resources! Carter’s teachers have even gone so far as to drop off additional materials at our door to engage Carter!

Kruse Family Christmas 2019.

So how do I incorporate all of this into our day? Making edible play-dough to teach measuring and baking cookies from-scratch to provide meaningful hands-on learning about following directions are great lessons. RIGHT?! On paper. But more likely, its refrigerated cookie dough, a slime kit daddy brought home, and a really great terrarium the Easter Bunny brought. The IPad is also our friend. Carter loves interactive games his teachers have recommended or we have discovered. It doesn’t take much to “allow” Avery to play an educational game on her phone! By afternoon, “academics” has taken its’ toll on the kids--we are all “done”--their brains and my non-teacher patience! We often try to get outside for some fresh air. This is a good distraction for Carter to break in his new shoes and braces. The kids love doing scavenger hunts or neighborhood lists of “things to look for”. On rainy days, I’ll be honest--Netflix is our friend! After dinner, a game of Sorry, Uno, or Candyland and its baths and bedtime.

COVID-19 has our nation all under a “new normal”. But as a cystinosis family, we live a different “normal” than most and because of that, we are no strangers to rapidly adjusting our schedules. The pandemic is requiring flexibility and accommodations from all and a “new normal” that is ever changing and we will continue to adjust. (However, I still have NO long term plans to homeschool!)

In Support of Rare Disease Patients Impacted by COVID-19, NORD Launches Premium and Limited Medical Relief Program

By Laura Mullen

Washington, DC, May 12, 2020 — United with more than 25 million Americans living with rare diseases, the National Organization for Rare Disorders (NORD®) today launched its COVID-19 Premium & Medical Relief Program to provide vital support to members of the rare disease community affected by the COVID-19 pandemic. The program will assist those with a confirmed rare disease diagnosis who have been directly impacted by the pandemic through a job loss, reduced work hours, furlough, quarantine, etc.

"COVID-19 poses significant threats to the rare disease community, given the susceptibility of those with chronic conditions and the economic challenges that affect their ability to weather this storm. We are hearing from patients who have been furloughed or laid off and desperately need financial support for insurance, COBRA payments, co-pays and medical expenses," said Pamela Gavin, Chief Strategy Officer for NORD. "Through this new program, we are providing premium and limited medical assistance to rare patients who need it most during this crisis."

NORD recognizes the needs of the community and has developed this program to provide critical support. The COVID-19 Premium & Medical Relief Program helps with certain out-of-pocket costs associated with health insurance premiums, and supplies eligible uninsured and under-insured patients with support for limited medical expenses. These expenses include medical visits and telehealth consults, laboratory and diagnostic testing, physical therapy and medical equipment.

We are grateful to Sanofi-Genzyme for their generous donation to this patient assistance program, which is available to all eligible rare disease patients affected by COVID-19. NORD has been proud to serve the rare disease community with assistance programs providing support to patients since 1987. In order to continue to meet the community's needs during this unprecedented time, NORD is seeking additional donations to its COVID-19 relief programs. Please help us to help our rare community.

For more information on NORD's COVID-19 patient assistance programs, including eligibility requirements, please visit our website, contact NORD by telephone at 203.242.0497, or via email at COVID19assistance@rarediseases.org. To stay informed, the NORD COVID-19 resource center presents regularly updated information and vetted links relevant to the rare community during this pandemic.

NORD®
National Organization for Rare Disorders

CRN Attends World*Symposium* Lysosomal Storage Disorder Conference, February 2020

Christy Greeley attended the World*Symposium* in Orlando, Florida in February 2020. This symposium is designed for basic, translational and clinical researchers, patient advocacy groups, clinicians, and all others who are interested in learning more about the latest discoveries related to lysosomal diseases and the clinical investigation of these advances.

A highlight was a symposium hosted by AVROBIO which included a detailed presentation from Dr. Stephanie Cherqui on the cystinosis gene therapy study. It also included presentations on gene therapy in two other diseases (Fabry and Gaucher) sponsored by AVROBIO utilizing the same platform. AVORBIO's experiences in these other areas are beneficial to cystinosis.

Anne Marie O'Dowd of Cystinosis Ireland, Dr. Stephanie Cherqui, and Christy Greeley at the AVROBIO symposium

AVROBIO Cystinosis Clinical Trial Update

At the American Society of Gene & Cell Therapy (ASGCT) Annual Meeting in May, AVROBIO presented new data on its investigational gene therapy programs for lysosomal disorders, including the investigator-led Phase 1/2 clinical trial in cystinosis.

New data from the first cystinosis patient treated with AVROBIO's investigational gene therapy (called AVR-RD-04) showed positive signs six months after treatment. Specifically, the patient experienced improvement in two key measurements of kidney function: estimated glomerular filtration rate (eGFR) and serum creatinine levels. There have been no reports of safety events attributed to AVR-RD-04 as of the last safety evaluation date (Jan. 27, 2020).

AVROBIO sincerely thanks the patients and families taking part in this very important clinical research, and the cystinosis community for its continued support.

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Leadiant Biosciences

ALAB Touch Base and Update

As we sit back and look toward the future, it is clear to see that there are many uncertainties in the world. One thing is certain, people possess and can build resilience during times like these. We all have the capacity to be there for one another, whether through singing happy birthday to a loved one from across the street, or just letting someone know you care by putting hearts on your window. The COVID-19 pandemic has hit everyone all over the world in a real, difficult, and continually changing manner. The community has been trying to navigate the path ahead with both their illness and newfound life or routine. There has been great fear, confusion, and uncertainty for the entire population during this time. We want you to know that we are here, we care, and we are always listening. So please reach out fellow Cystinosis Warriors, reach us on our Adult Leadership Advisory Board (ALAB) Facebook page, direct message, email during one of the video sessions, or in whatever way you feel comfortable.

Cystinosis Rare: A Journey into the Unknown Podcast

ALAB's quarterly podcasts hosted by Cheryl Simoens, Steve Scheudler, Jana Healy, and Sara Healy focuses on tackling questions that were developed by both the hosts and community members in a discussion and Q & A format with many experts and medical professionals.

The first episode focused on Success Stories from the hosts and two adults within the community. It was inspiring and humbling to hear from fellow Cystinosis Warriors on their successes in life. The second episode focused on grief, loss, and mental health. This episode dealt with issues on loss of abilities, voice, or any loss that the illness has brought throughout our journey. The third upcoming episode will be released in August and will focus on transitioning from youth to adult care and the challenges that arise. We would love to hear from the Cystinosis community on their experiences and what advice we all may have for the younger generations or questions community members might have on transitioning. Please reach out and contact us through our ALAB Facebook page.

Cystinosis Sessions

Cystinosis Video Sessions is a platform for members of the community to join virtually and discuss topics facilitated by ALAB members Brian Ensor, Eddie Jr. Langley, Emily Mello and Karen Gledhill. The first session included the topic of medication with a discussion over how we take medications, everyday routines and addressing odor. The second session focused on self-reliance, education, and employment and the upcoming episode in June will focus on facing challenges, peer pressure, limitations and overcoming them. Stay tuned!

CystinosisTEENS Instagram Account

CystinosisTEENS is a private Instagram account for teens between the ages of 14 and 21. It is a space for teens to discuss, learn and connect in a safe and friendly environment.

The ALAB was well-represented during the 2020 Rare Disease Day events. See more on Page 30.

2020 In-Person Meetings Are Out, Customized Virtual Meetings Are In!

This year we'd prepared a series of cystinosis regional meet ups around the U.S. Due to COVID-19, we've made the decision to cancel those in-person events. But living with cystinosis isn't on pause while we deal with the pandemic; we know how much it means to shake hands, converse, laugh, cry, and hug with someone who really gets this disease. We are determined to continue connecting individuals and families like yours.

Our solution: virtual meet ups beginning this summer. Perks of moving the events online include:

- **Accessibility.** Not everyone is able to travel
- **Less Geographical Restrictions**
- **Flexibility.** Choose the meet up date/time/topics most appealing to you
- **Customization.** This series is being designed exclusively for our cystinosis community
- **Staying Safe!**

Event information is being updated regularly. Please check our social media channels and website at cystinosis.org. This summer's virtual meet ups are going to be a unique experience. We hope you can join us!

ALAB Poster Presentation
National Kidney Foundation Virtual Event

Maya Doyle, Karen Gledhill and Eddie Langley Jr. attended the National Kidney Foundation (NKF) virtual event in March to present a new poster developed to advocate and promote the Adult Leadership Advisory Board on who we are, goals, programs, and services we provide along with our mission.

Cystinosis Awareness Day 'Succeeding with Cystinosis' Challenge

The entire Adult Leadership Advisory Board (ALAB) participated as a collective group in our new 'Succeeding with Cystinosis' Challenge displaying images of how they succeed. We also asked the community to show how you 'Succeed with Cystinosis' through a photo, description or image. Boy did we get a huge response! We received photos of community members and their beloved family members participating in their favorite activity or hobby, engaging in physical activities, and educational accomplishments. Sara Healy successfully turned the photos into several collages that were displayed on May 7th to commemorate Cystinosis Awareness Day on our ALAB Facebook page.

Eddie Langley Jr. and Emily Mello created a special tribute slideshow with pictures of all the Cystinosis warriors who had passed along the way. You can view the slideshow at https://bit.ly/2Ypw0lp.

Family Support Update

By Jen Wyman, Vice President of Family Support

These are strange times. Living in the time of a pandemic is scary and there is so much unknown, but in many ways we, as a "disease centered" organization, are all too familiar with what it takes to stay healthy… social distancing, wearing masks, washing hands, and sanitizing. These are all things we do when we have immune compromised people in our lives. These are all things that those who have experienced transplants have already done. So while this is a time of unrest and unease, we as a community know how to handle it. This too shall pass with planning and patience. We thrive under stress. We've got this.

The Wyman Family Summer 2019.

Sadly, we have lost our fair share of cystinosis warriors this year and at the same time have welcomed newly diagnosed families. Cystinosis knows no boundaries and it gives and takes as it pleases. When you live with cystinosis you learn to live with some level of uncertainty. You journey into unchartered territory on a daily basis. You understand that you have to give up some control in order to maintain sanity. We also know that our dark days don't persist. They come in waves…sometimes the darkness lasts too long, but it doesn't last forever. This too shall pass with hope and humanity. We thrive under pressure. We've got this.

Jen and Kacy Wyman show off their masks.

Idea to Reality – A Tribute to the Cystinosis Warriors We Have Lost

By Eddie Langley, Jr.

Cystinosis is what it is… a lifetime adventure full of victories, defeats, pain, relief, miracles and loss. There's just so many variables when you live with this horrible disease. Through my 41 years of life I have met many people who share this rare disease; some seemed to struggle with the different side effects more severely and others, not so much. Before social media I remember meeting maybe 2 other kids with cystinosis. I told myself I never wanted to hang out or befriend another person with Cystinosis because there was so much anger and negativity. Then through the years my mind would start to wonder and I would think to myself; maybe they lost a sibling like I had, how many procedures did they have to endure, or maybe they were having trouble with bullying and did not know how to deal with it. There's just so many things out there! Not being old enough to understand the things my sister went through I would start to think about her even more. I began thinking about others going through all the trials and tribulations. I have 2 memories of my sister; one was wanting some food off her hospital tray, the second was looking down at her from the arms from one of my parents and saying goodbye to her at the viewing. The older I became the more I would understand my previous encounters. When social media came out I started to meet others with cystinosis and even more parents. The more I got know everyone and getting to know their stories I came up with an idea to remember and honor those that have passed, but I had no idea how. As the years went on and technology became more sophisticated and user friendly my idea came to me about having a slide show and having music along with their birth and death day. Over the years I have seen so many improvements in the treatments available to our small community, but one thing has not changed, the struggles we all face. My decision was made after trying to put my tribute idea out there for a project with no luck. I pulled the trigger having experience in playing Oregon Trail from the 80's and Microsoft Word for computer experience and my idea became a reality.

To view this touching tribute, please visit https://bit.ly/2XY0URs.

Tribute to Robert "Robbie" Smith

By Emily Mello

Robert Eric Smith was born on July 14,1979 to the most wonderful, kind hearted, bright spirited parents I know, Debbie and Robert Smith. Their so loved and precious first born came into the world to make them proud, first time parents. He was a gorgeous, blue eyed, baby boy who soon showed signs of excessive thirst and swollen wrists and knees that needed medical attention, like so many similar stories we hear. They were worried and desperate for answers to why their little boy was so ill, jumping from doctor to doctor, being told that he would not see his 5th birthday. Some of their questions were finally answered when at the age of 4 their precious gift was diagnosed with Cystinosis at Ormond Street Hospital in the UK.

Their baby boy and the family were now presented with new challenges, a new diagnosis that most doctors had never heard of, frequent hospital stays and visits, new routines and medications, including the dreaded, yet necessary Cystagon, to keep their baby alive. They now had their diagnosis and the tools for their baby to grow and lead a normal life. The greatest tool they had was the love and support of each other and their family.

Time passed, Robert was growing and became a big brother twice-to Sonia Smith and Sean Smith forming a beautiful family of 5. Whatever name you knew him by (Robert, little Robert, Rob, Bob, Bobby, Robbie, babe, or honey) you knew how strong he was. Rob grew up in Maidstone, Kent UK with his beautiful family always helping him through his journey in any way possible.

He made his way through school. He was hard working and had many demanding jobs, something that I have always admired. Rob did a carpentry apprentice and went into graphics with his brother Sean. They did some graphics billboards and posters for different businesses, including one for the National British Museum that I had a chance to visit with him and see his work on display. I remember thinking wow how amazing his work was and I felt proud standing in front of his work. Rob had his first kidney transplant at 16, donated by his hero mum, Debbie. He was able to accomplish so much with his new kidney. A few years later, after spending 4 years on dialysis and a scary moment in ICU, he received a second kidney transplant at 25 years old. Rob was always a person who persevered, who kept going, kept fighting, kept learning, kept setting goals and achieving them, never allowing cystinosis to take over. It tested his limits over and over, but he never complained; He just did what he needed to do and got it done. When Rob was 30 he passed his HGV Lorry test and became a Lorry (truck driver) for the NHS, delivering hospital equipment, a job I know he took pride in. He was always driven to his do his best in this job. He had early morning shifts, night shifts, and long shifts that took a toll on him, but he did it and he did it well. Our time difference had

us up late hours and I can still hear him telling me how he was feeling “knackered’ from work.

Robbie and I met on a trip my son, Elijah, and I took in 2013 to the UK. We were both part of an online group for adults living with cystinosis. I had mentioned to the group that we would be in the UK and if anyone would like to meet us please call. Robbie came to meet us. April 4th, 2013 will forever mark our story together. This is the day Robbie came into our lives and changed it for the better. We met in the Tavistock Hotel lobby in London where he immediately befriended Elijah. They had a special bond. He was patient and loved Elijah.

The next two days were very special. He took us touring in London. It was then that Robert started making my dreams come true; he just didn’t know it yet. Those two days were enough for me to want to be more than just friends with this amazing human being. Cystinosis brought us together and all I wanted was for the day not to end. Sadly we had to come home back to America, but we kept in touch. We spoke every day, we felt like teenagers talking to each other all night, not wanting to hang up, everyday we would make each others days by texting and soon we became accustomed to this. We wanted more. He became “our Robbie”, as Elijah would say. Rob planned his first visit to America with his brother to New York. We toured New York and we made our own memories that will always be apart of us and can never be taken away. Robbie and his brother Sean returned to the UK, but this time I was different. Robbie and I decided to be a couple (we even made it Facebook official). Over the next five years we traveled back and forth to each other. Our little family of three saw the world together and created lasting memories. He made so many of my dreams come true. He was a wonderful and caring boyfriend and a patient and amazing dad to Elijah while we were together, and our extended families were tied together.

Every painful goodbye at the airport became too hard for us to keep going. Airline ticket prices were rising, work and life schedules made it harder and harder to maintain our long distance relationship and we agreed to come to an end… Rob there in England and me here in USA. I believe that our care for each other never went away from the time we met until his

last breath. My love for Robbie is will never end. We lived so passionately while together, as a family, as a couple, and as best friends. Our love and respect for each other was immeasurable. His heart was so big and his kindness, compassion and generosity was endless. He was the most amazing man in my life and we loved each other unconditionally. I had the pleasure of being “his” for awhile.

I can not give a tribute to Robbie and not mention his beloved dog, Fifi, who is now 17 years old. She was his companion and he took such good care of her.

Robbie lived a full life. He left this earth at 40 years old on April 22, 2020. Their is immeasurable pain in my heart knowing I will never hear from him again, but I find comfort in our memories.

In honor of the man I fell in love with, to his family that I share my pain with, may Robbie always be remembered for the amazing man he was.

Robbie is joining his granddad Robert Smith, Nan Josephine Smith, and cousins Paul and David in paradise. He will be missed by his mom and dad Debbie and Robert Smith, sister Sonia Smith, brother-in-law Jamie, nephews Kian and Ethan, niece Lilly, Brother Sean, sister-in-law Josie, nephew Sebastian, Fifi his companion, many uncles, aunts and friends and so many cystinosis warriors who had the pleasure of knowing Robbie.

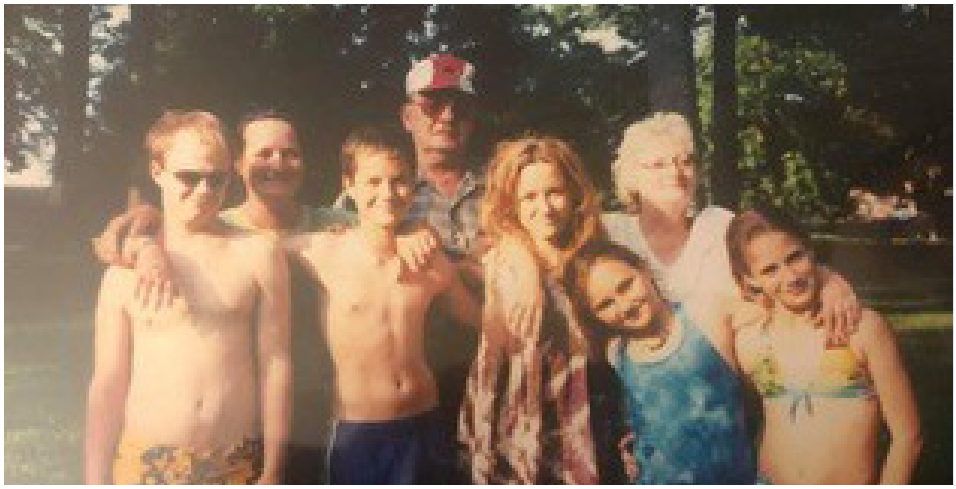
A Tribute to our Big Brother — Todd Bradley

By Emily Mello

I first heard of Todd before I actually met him. I had heard of this ambitious guy who was passionate about helping all who were going through this life with cystinosis. I heard of his many ideas and plans he had for the community. Both of us took part in the early stages of a project called Future By Design, of which Todd eventually became president. When I finally had the chance to meet Todd at the Cystinosis family conference in Utah 2017, instead of a handshake I was welcomed with a warm big brother hug from a guy I had never met before. I remember his green hat on backwards, and warm smile; I said “so your Mr. Bradley? I’ve heard a lot about you.” He replied with a laugh and a yes I’m Todd, nice to meet you Em.” At that moment I knew this guy was going to be a good friend.

We kept touch after the conference, became friends on Facebook and looked forward to what was ahead for the project, Future By Design. It was in late November, 2017 that we both had the chance to travel to Portland, Oregon for a Future By Design summit with other great adults living

with cystinosis. I had the chance to be part of a smaller group with Todd to come up with ideas on how to help our community cope with cystinosis and its challenges. It was there a dream grew in Todd's heart to start a YouTube channel speaking openly about cystinosis and its challenges. "Talking with Todd" came to life and his many videos are posted there. I encourage everyone to watch. We had the pleasure of working with adults with cystinosis and had the chance to get to know each other. It was an opportunity that Todd was so grateful for. He made a great impact in the community. So many memories were made during our time in Portland. Shortly thereafter our beloved Rachel Patch passed away. I can remember like it was yesterday, calling Todd to ask if he heard about Rachel, and just crying together on the phone. We didn't say anything to each other, we just cried and mourned the loss of our friend. It was from there that our friendship grew. He would check on me, making sure I was OK. He protected me and made me laugh. He was like my big brother, always smart with his remarks, always making a joke and always bringing my spirits up. We would see each other in smaller meetings and listening to Todd speak about his dreams and his own story made me so happy to have



a friend like him. Todd was all about including everyone, making them feel welcome, and if you needed a friend, he was there. The last time we saw each other was at the latest Cystinosis conference in Pennsylvania. He introduced me to so many beautiful people and I saw the passion he had for them, and how good he felt that he was a part of helping others. His heart was big. When we said our goodbyes and shared an extra long hug, little did we know this would be our last shared hug.

He always encouraged my son, Elijah, to do well in baseball and in school. We disagreed on football teams and that became a way we stayed connected-always texting each other who the better team was. He was an Eagles fan. He would tease my son about this, as he knew Elijah was passionate about the Patriots. I would call him Sir, he hated it! So I continued calling him Sir. He was fun to banter with.

We kept in touch and he became my "go to guy". I trusted him with everything I said, and he trusted me. He was the big brother I never had. I can ask around and know that others have felt the same way. Todd was a mentor, a counselor a great listener. He was funny and always available to lean on.

"Merry Christmas Em!" was one of the last texts I received from Todd. It followed a phone call just two weeks before letting me know he was ok. The conversations were many and ones that I will hold dear to my heart. Todd was a dreamer. He set goals and he wanted to help others. He wanted to make an impact in someone's life and he was worried that he would not have accomplished this. I assure you, he did! He impacted mine, and so many more lives, with his videos, with his care. I know how much he loved his family, and the cystinosis community was very important to him. He will leave an emptiness in his family's lives, and in this community and especially in mine.

Forever in our hearts Todd Bradley,

We will miss you, Sir.

Chandler Moore mans the raffle table.

50/50 Raffle

Feeling the love on a local level. Our friends at NWTF Lower Delaware held their annual 50/50 + raffle in February benefiting the CRN. Our sincerest thanks to the organizers and donors.

FAMILY STORY:
The Sevels

By Mike and Kristina Sevel

Mike, Grace and Kristina Sevel.

Grace Sevel.

Early May of this year we called our pediatrician after noticing that our daughter Grace had been vomiting during car trips, and having seen some shaking of her hands after waking up from naps. COVID quarantine was still in full swing so we took a short video of Grace's hands and had a telemedicine visit with our pediatrician. After a long conversation, and her observing Grace and asking us to do a few different tests, our pediatrician said that she wasn't concerned but we should order some routine labs just in case. I loaded Grace in the car, went for the labs and expected to hear from the pediatrician the next morning. At the end of the business day she called to express that Grace's labs were abnormal and recommended we go to the emergency room at Rainbow Babies & Children's hospital. She said it could have been a bad blood draw but didn't feel comfortable waiting until the next morning to have the labs redrawn in the outpatient setting. After a few hours in the emergency room we were told that Grace's labs were accurate and they believed she had something called proximal renal tubular acidosis. They did not know why but that we would be staying overnight to correct her blood chemistry imbalance and we would work with genetics after we were discharged to determine the cause of the RTA. After a long weekend which included a transfer to the PICU a new nephrologist took over Grace's care Monday morning. She informed us that blood work had already been drawn and was on its way to California because she suspected that Grace had a disease called nephropathic cystinosis. Up until this point, Grace had been a completely "normal" healthy baby. She had slowed down on the growth curve but our pediatrician just believed that she would be on the small side. The 4 days that we spent waiting to hear about a confirmation of cystinosis seemed endless. Due to the COVID pandemic we were unable to have any visitors and it made the possibility of such a rare diagnosis even more isolating, but we focused on healing our baby and soaking up all of the information that we could. Friday evening Grace's nephrologist sat down and while Grace slept in my lap confirmed that Grace's WBC cystine levels were elevated, therefore confirming her diagnosis. She expressed how wonderful the cystinosis family was and that while this would be difficult we would have a wealth of support from other families who were in our shoes. We didn't know just how true that would be. We spent another 13 days in the hospital for a total of 21 before finally coming home. After connecting with so many families and reading all of the information available we advocated for Grace to have a G tube placed prior to our discharge, it was the best thing we could have done. We are so thankful to finally be at home, learning how to care for Grace and ourselves with the support of our family and friends. We live in a small suburb east of Cleveland, Ohio with Grace's best friend, our German Shepherd, Carlo. Thank you from the very bottom of our hearts to this community for being so strong, we appreciate all of the work you have done thus far and cannot wait to get to know all of you further.

Mike and Grace.

Grace.

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.

Cystinosis Research Network and Amazon Smile

Support our organization while you do your holiday shopping on Amazon! Designate CRN as your favorite charity on Amazon and 0.5% will come back to the cystinosis community.

How do you do it?

1. Visit smile.amazon.com
2. Select your account
3. Under "Settings" select "Change Your Charity"
4. Type and select "Cystinosis Research Network"

amazonsmile
You shop. Amazon gives.

Jack Greeley 20th Birthday Fundraiser for CRN

By Christy Greeley, Vice President of Research and Executive Director

St. Patrick's Day 2019 in Chicago

The Greeley Family once again celebrated Jack's St. Patrick's Day birthday by sending out our 18th edition of his birthday fundraising letter. This year of course was quite different than most given the stay at home orders in place, but as usual, Jack made the most of it. We are so thankful to our friends and family who continue to support not only our family, but also the greater cystinosis community – Jack's birthday fundraising total over the years is approaching the $500,000 mark, a blessing indeed. Following is an excerpt from this year's letter:

We are writing this letter on Jack's actual birthday; #20! This year he and Christy have spent A LOT of quality time together social distancing in the house during the COVID-19 quarantine. This is the complete opposite of a typical St. Patrick's Day birthday celebration for our family. Last year, for example, we spent March 17th at the Big 10 tournament watching Michigan play and celebrating the dying of the Chicago River. The slowdown of day-to-day life with the coronavirus pandemic turns out to be a chance to gather thoughts and put together our family's annual letter celebrating Jack and asking for your help in supporting our fight for the Cystinosis community. And, scoop, this is Christy's first stab at being the lead author, so here goes.

For 18 years now, we have been updating our "circle" about our son, Jack who on many levels is a marvel in his own right whether past, present, or future. As most of you know, Jack has cystinosis, a rare metabolic, genetic disease affecting about around 600 people in the U.S. and around 2,000 people worldwide. Christy has now served nearly 18 years as Executive Director of the Cystinosis Research Network (CRN), an internationally recognized 501(c)3 whose vision is the acceleration of the discovery of a cure, development of improved treatments and enhancement of quality of life for those with it. Cystinosis is a genetic disease typically manifesting during the first year of life as progressive kidney disease resulting in renal failure during childhood, if not treated. In cystinosis, the metabolism of the amino acid cystine is defective leading to its accumulation in the kidney and other organs. This cystine accumulation results in cellular damage and death, but the direct mechanisms beyond this phenomenon are largely unknown.

While that sounds scary, the Cystinosis community has come a very long way in the 19 years since Jack's diagnosis and there are many reasons for hope and optimism. Some examples include:

- Two new treatments approved by the FDA are now available; Cystaran, an eye drop which dissolves crystals in the cornea not treated by systemic medication, and Procysbi, an extended release formulation of the existing treatment, Cystagon.
- Gene therapy studies which may eventually have the power to halt the progression of the disease have begun enrolling patients with early, promising results.
- Patient advocacy organizations like CRN have been able to utilize social media and other online tools to reach out to families with education and support much earlier, with more efficiency and more powerful and accurate information than ever before. This enables families to connect, to receive better treatment, and to ultimately create a better, healthier life for individuals living with this disease.
- Adults with cystinosis have benefited from better treatments and are living longer, healthier lives. They have become active advocates within

our community, with their own voices to share and contributions, particularly evident in the activity of the recently formed CRN Adult Leadership Advisory Board. Check out all they have accomplished on the CRN website at https://www.cystinosis.org/support-resources/alab/.

Jack has himself spent these last 19 years in many ways like every other kid growing up, but in others engaged in the most extraordinarily impressive battle against this disease. When Jack was diagnosed around his first birthday in spring 2001, he became positioned for a lifetime of picking, poking, prodding, nagging, cutting and examining. A chronic disease forces a patient to have discipline and vigilance tied to medical routines that were previously taken for granted. For Jack, this has totaled nearly 200,000 pills swallowed (divided up every six hours), including getting up in the middle of the night, for nearly 7,000 consecutive days. There have been around 60,000 eye drop applications and well over 500 medical appointments. Of note, Jack has done pretty well on his own managing this at college. He has also faced surgery five times so that doctors could address ongoing issues:

- Once to insert a gastric feeding tube into his stomach because he did not eat solid food for 18 months;
- Twice for knee and ankle surgeries to help straighten these body parts with staples and screws;
- And twice for highly invasive orthopedic surgeries to help straighten his lower left leg. One included a radical, complicated set of procedures in 2017: Triple C procedure (three bones in his left foot were surgically cut and shifted with cadaveric wedges inserted to straighten the foot), de-rotational osteotomy of the tibia (the tibia and fibula bones were cut and rotated to correct alignment and then secured with a metal plate and screws), and installation of an eight plate (his left knee had another eight plate inserted to take advantage of any potential growth remaining to further straighten the leg).

The most normal and typical accomplishments always seem more extraordinary for Jack, though in most ways his life is like any 20-year-old. He is finishing up his sophomore year at Marquette University, where he is busy majoring in Anthropology and competing with the Marquette ESports team. In fact, he travelled on his own last month to Montreal to join teammates at an ESports tournament there – a great step towards any young adult's independence. Jack loves exploring Milwaukee, and is particularly adept at finding the most amazing variety of restaurants for us to try out when we visit. Let him know if you need any recommendations.

Although our family has changed a bit these days, with Dave working in Austin, Alex moving to Nashville last October to begin her first post-graduation job, Jack studying in Milwaukee, and Christy living in the Chicago area, we continue to work together to love and support Jack. We want to do our part to ensure the Cystinosis community continues to grow and address the needs of individuals like our Jack. Please consider supporting him and our efforts with CRN by making a donation in his honor and his 20th birthday this St. Patrick's Day. Thank you, God Bless, and we wish you and your family well.

Marquette Family Weekend 2019

Jack's ESports Team enjoying a Marquette basketball game

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.

goodsearch

Search the Web with GoodSearch & Raise Money for CRN

Every time you use GoodSearch.com to search the Web, a donation is made to the Cystinosis Research Network! GoodSearch.com is powered by Yahoo!, so you get the same results you get from most search engines. What is unique is that GoodSearch.com has developed a way to direct money to your selected charity with every click!

To get started, go to GoodSearch.com and select Cystinosis Research Network where it says "Choose your cause." Then search like you normally would!

The more people who use this site for CRN, the more money is earned. So please tell your friends and family!

goodshop

Make Purchases at GoodShop & Raise Money for CRN

GoodShop.com allows you to purchase through most online retailers, and a percentage of your purchase goes to the Cystinosis Research Network! There is no additional cost to you!

To get started, go to GoodShop.com, choose CRN as your charity, and click through the link on the GoodShop page to get to your favorite retailer. It's that easy!

GoodShop will donate up to 30% of your purchase to CRN. Some of the hundreds of retailers include: Best Buy, iTunes, Home Depot, Amazon, Barnes & Noble, Dell, Banana Republic, Macy's, Target, Wal-Mart, Ann Taylor Loft, Chicos, Coldwater Creek, American Eagle Outfitters, and many more!

2019-2020 Livgracefully Updates

By Kirsten Stilke

For those of you who are not familiar with Livgracefully, let me take a moment to explain the story behind our mission.

Both of our children, Mason and Livia were diagnosed with Cystinosis. In 2014, at 8 years old, Livia suddenly and unexpectedly passed away. She did not pass from Cystinosis, but rather a different rare genetic disorder called Fibromuscular Dysplasia. We had absolutely no clue that Livia had this other disorder, so you can imagine when our seemingly “healthy” child passes away; it leaves a devastation one cannot explain.

Livgracefully was an idea our family had to not only honor Livia, but to share her story with others. Our loving and supportive family had designed the logo and had helped us form our nonprofit! Our mission in Livgracefully is to give back to our community in the form of local monetary donations and participate in the local Scholarship program. We also give back to the Cystinosis Research Network and Fibromuscular Dysplasia Society of America. Livgracefully is a 501c(3) that is solely run off donations from generous supporters like you!

Since its inception in 2017, Livgracefully has not only helped so many people and organizations but, it allows us to not only honor her memory, but to pass along what she PROFOUNDLY taught us about life.

Although it was NOT cystinosis that took her from us, it is that which allowed us to learn from both Mason and Livia. They bravely fought each and everyday against the disease and courageously put up with more than most adults would have.

As we know, health issues and diagnosis are devastating--now imagine you are a kid, trying to figure out the world and also have this health crisis to deal with. Livgracefully is a way for us to acknowledge kids who faced similar challenges; yet hold onto the hope that these obstacles will not stop them from achieving their goals. Often times; we don’t realize what a child or even an adult is going through. Despite these private struggles, these kids teach us what it means to have faith and continue to be brave whatever health situation they may be dealing with. Our kids know what that battle feels like and continue to persevere; even though at times it is difficult. We have chosen to honor Livia’s memory by recognizing students in similar situations. We can no longer send her to college, so we’ve decided to make it our mission to help someone else achieve their dreams!

Each year, Livgracerfully’s main mission is to award two $500 scholarships out to local Elkhorn High School Seniors who share their stories with us; their struggles, yet shared their dreams and hopes and how their

adversity with health issues helped shape them; not letting it define who they are.

We have received anywhere from 25-60 applicants each year! It is outstanding the number of kids who are going through these battles that we don't always see; and how they persevere is astounding! Our family and our Board of Directors sift through each essay; which is such a difficult task to try and only pick 2. Each year we wish we could recognize each and every person who applied and give all of them scholarships!!

2020 marks the THIRD year we have given these scholarships; which added 6 high school seniors to the Livgracefully Family! We are overjoyed at the mere fact we get to do this! And we hope to continue to do this for a very long time!!

Each year, we are often found running a Facebook (Network for Good) campaign and rely on Amazon Smile donations to help us reach our goals!

We have successfully sold apparel, bracelets and stickers to help raise money for our cause! We are hoping to soon host a Run/walk or Golf Tournament fundraiser locally to help allow us to not only get bigger; but to help us be able to give back MORE!!

This year, we will be launching another Livgracefully apparel line!!! We are hoping with the current health crisis, we will be able to launch at the end of summer/early fall!

Recently, Livgracefully has had the pleasure of being able to donate $1000 to CRN! We also donated to FMD Society of America, Make-A-Wish and of course the coveted local pet shelter Livia loved; Lakeland Animal Shelter. Our Livgracefully Scholarship recipients will be awarded their scholarships on June 3rd, 2020 in a virtual award ceremony-due to the current pandemic situation.

Thank you for allowing us to share our passion in this life and our ongoing mission to honor Livia and recognize our kids, whether they have cystinosis or not; who teaches us daily what it means to persevere against the adversity life has thrown at us!

We invite you to follow us on Facebook (Livgracefully) and our website livgracefully.org for the latest up to date fundraising information and just to check in and see what is new with our group!

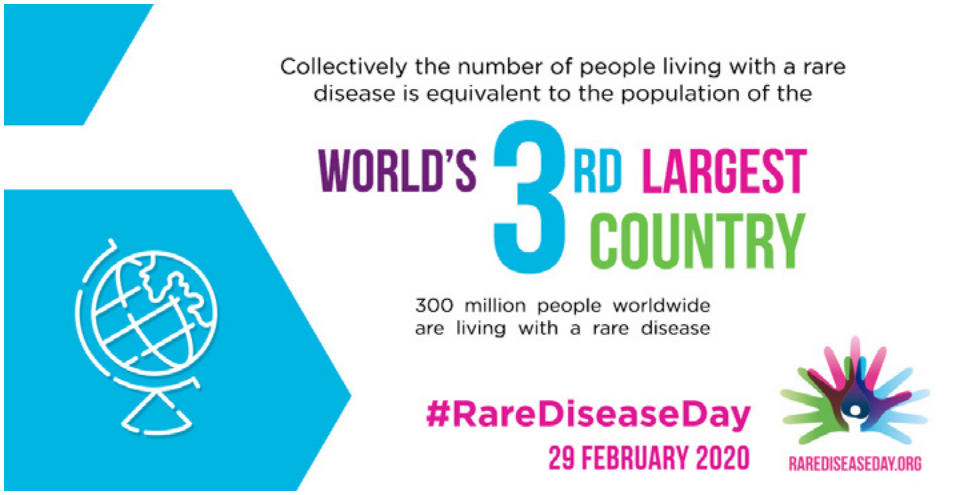
CRN Commemorates Rare Disease Day

February 29, 2020

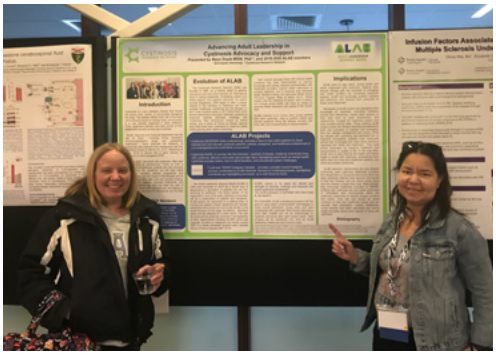
This year Rare Disease Day fell on Leap Day making it one of the rarest of rare. We wanted to commemorate the occasion by proving a simple way to raise awareness throughout the year. Leading up to February 29th, the CRN supplied free phone wallets to all who signed up. Our awareness ribbon was customized to stand out against a bright green background so we could “share our rare” whenever our mobile devices were around.

The aim was to spark conversations about cystinosis and the underserved rare disease population. Through the campaign, the phone wallets traveled to 13 countries spreading awareness, education, and love for our cystinosis warriors. Thank you for supporting this program!

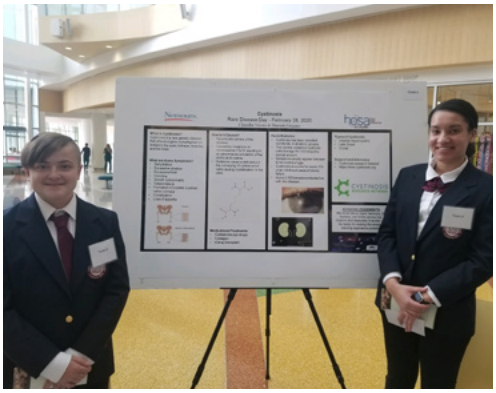
Did you know? The inspiration behind the adapted ribbon image was drawn from a 12 foot (3.66m) tall structure; meticulously created using 2,000 interior lights, one light for each person living with cystinosis. The ribbon was outlined in a contrasting color to represent those who have passed but remain part of who we are today.



Advocacy in Action: Rare Disease Day



Janice Finn and Emily Mello at Quinnipiac University.



Chandler Moore (left) at Nemours/A.I. DuPont Hospital for Children.

Sara and Jana Healy at University of Minnesota.

Jenn Loglisci at Bay Path University.

Our cystinosis advocates showed up to ensure the cystinosis voice was heard this Rare Disease Day. In addition to the Rare Disease Week lobbying on Capitol Hill, we'd like to recognize the efforts of these individuals. Thank you to all who participated!

Hamden, Connecticut

Emily Mello and Janice Finn were Rare Disease Ambassador's at Quinnipiac University's 5th Annual Rare Disease Day symposium in February 2020 (more event info available here: https://bit.ly/2YtRDkq). Accompanied by Maya Doyle, a professor of social work at QU and longtime cystinosis advocate, they (also) presented the poster describing the mission and activities of the Adult Leadership Advocacy Board (ALAB). In addition, they hosted a table with CRN materials and spoke with medical students and fellow advocates about living with cystinosis and rare conditions. This year, many of the speakers and panelists at Rare Disease Day focused on the work of inclusion for individuals with disabilities and chronic health conditions across a wide spectrum of rare disorders.

Wilmington, Delaware

Chandler Moore represented the cystinosis teens with a presentation at Nemours/Alfred I. DuPont Hospital for Children.

Duluth, Minnesota

Sara and Jana Healy attended a Rare Disease Day event at the University of Minnesota. The keynote speaker was Sarah Wicks, JD, MPH. Her focus is on helping patients and rare advocacy groups become more involved with rare drug development processes and giving a voice to the community through sharing everyday experiences. She recognized how personal experiences could help with drug development. She discussed the process of setting up an FDA meeting to share stories, explain what is required, and how to get the drug approved. She helps rare disease communities with the whole FDA process.

There was also a discussion on the Minnesota Rare Disease Advisory Council with Erica Barnes, RDAC Administrator. Their focus is improving care for the rare community and developing recommendations and resources to improve access to and coordination of care. Erica Barnes also heads up the Rare Action Network of Minnesota. She is an amazing advocate for the rare disease community. It was a great experience and we look forward to the event next year. The duo presented work in progress by the ALAB.

Longmeadow, Massachusetts

Bay Path University invited alumni Jenn Loglisci to speak during Rare Disease Day. Feedback from students was overwhelmingly positive and they valued hearing Jenn's point of view.

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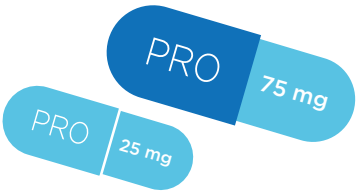
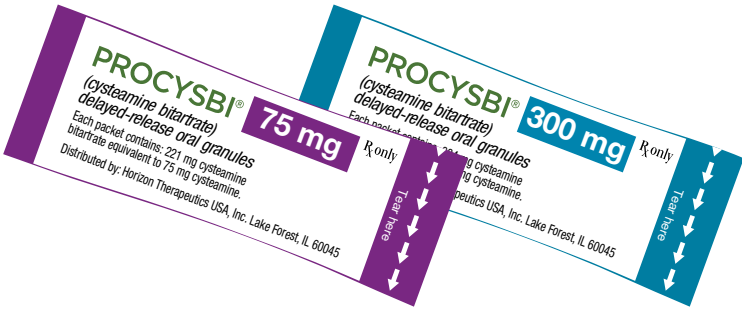


Available packet strengths:
75 mg and 300 mg



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- Provide education and answer your questions

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- **Visit PROCYSBI.com**

HORIZON

Rare Disease Week

By Clinton Moore, President

Rare Disease Week in DC is a very inspiring week to say the least. If you can imagine hundreds of rare disease patients, caregivers, and organizations all in one place at the same time with the same exact thing in mind can be quite inspiring. Really puts into perspective that we are not alone. Not even close.

This year I was joined by four others from our community. Jonathan Dicks, Melanie Vachon, and Leslie and Tina Cogar. As it was their first time attending this week long event, I could see the same concerns and worries that I had the first year I attended. These concerns were quickly left behind as the week began.

As we did have time to relax and enjoy each other's company in the evenings, the conversations often led to the same comments. As we would discuss the day's events, the four that joined me would express that they really felt they were making a difference and had no idea that so much for rare diseases was happening in the nation's capital. My response was always the same.... they were making a difference. Even if you don't see a change right away or directly for the cystinosis community, they needed to keep in mind that many improvements have been made on a legislative level that benefit the rare disease world as a whole, all because of people like them taking the time to make their voices heard.

Talking to your state's representatives or congressmen and women can be nerve wracking at first, but just like myself they told me that after a few minutes they realized that those that they were talking to were humans too and noticed they were taking a lot of notes. They said they felt that the representatives really cared. That's because they do. They listen. They have no idea of the problems or concerns a community is having unless someone steps up and tells them. That's how changes begin to happen.

I can't thank Jonathon, Melanie, Leslie and Tina enough for joining me for the week. Taking time away from your families and lives is a lot to ask but you didn't think twice. Thank you for representing the cystinosis community and the Cystinosis Research Network. Your efforts make a difference in the lives of all rare disease patients, not just the cystinosis community.

Rare Disease Week: An Interview with Jay Dicks

Advocating for rare disease sometimes feels like fighting an uphill battle. Living with cystinosis means you are 1 in 2,000 people in the world with this condition. Each year during Rare Disease Week, the CRN makes the journey to Washington, D.C. to fight for the rights of our cystinosis warriors.

Elle is four years old. Her father, Jay, attended his first-ever cystinosis family conference in July 2019. After promptly forming bonds with other individuals and families with cystinosis, he knew he wanted to be more involved. As one of the cystinosis representatives on Capitol Hill during Rare Disease Week, here is some insight into his personal experience – advocating for his daughter's rare disease.

CRN: What were your expectations for Rare Disease Week?

Jay: My expectations were that we would be coming together with the entire rare disease community for the common goal of raising awareness. I do not think I was ready for the sheer size of is aggregate, it was awesome to see and at first a bit overwhelming. I expected to sit in some meetings and talk about policy. I was pleasantly surprised that we were tasked with "owning" our time with our elected officials and actually got real time to sit down and talk with these incredible people.

CRN: What did you think of the experience?

Jay: It was life-changing for me as a parent with a child who had been newly diagnosed. There is a feeling that no one outside of your specific rare disease community really understands what you go through. Family and friends are well-meaning but there is a void that exists nonetheless and traversing the day to day struggles of living in a world that is not tailored to your unique needs has a way of feeling like you and yours exist on a deserted island. This trip put so much in perspective for me. To say I was humbled is an understatement.

Our representatives from House and Senate were so kind, understanding and respectful of our time. At the core of all this what spoke to me was that what we were in fact advocating for was common sense legislation with regards to the rare disease community, nothing more. When I realized this, the advocacy was so easy to translate from my micro-community in Cystinosis to the macro-level of a national stage.

CRN: What was your key takeaway?

Jay: Getting the wheels of "change" turning from the policy levels takes a concerted and systematic effort on many levels. One also needs to understand that diligence and patience are needed as the process is not something that comes about immediately. Stay the course, do not be discouraged. Emotion is great, but composure is paramount above all else.

If I cannot effectively get my story and reason for attending across the table without turning into a messy puddle of snot and tears, then what did I really accomplish? Sympathy, is not

what I wanted to personally promote, empathy was the goal. Empathy begets action behind it, sympathy seems to gum the wheels of change and mires us in a perpetual cycle of self-pity.

CRN: Would you do it again? What would you say to someone considering this level of involvement?

Jay: I would do this again in a heartbeat. The advocacy at this level is all about having your "why" concisely planned, and to be able to hit the major "talking points" without going too far into the minutia of the disease itself. The major takeaways for me are:

Practice and practice the "elevator speech".

Your representatives in the House and Senate represent you, you elected them, so they are on your side, regardless of past voting history

Therefore, go into the meetings without preconceptions, you may be pleasantly surprised.

Get to know advocates from other sectors of the rare disease community.

Some are veterans and have incredible knowledge, tap into that and you will find like I did that the community you "think" supports you is much larger than you thought.

Be a sponge and be humble but speak with conviction. Get business cards made. So much is happening in such a short time that it is impossible to remember everyone you met. Pass yours out in abundance and ask for others with reckless abandon. Follow-up with everyone.

CRN: What has being on Capitol Hill this week meant to you?

Jay: This week meant that I have a much larger voice and audience than I ever expected. My 4 year old daughter's voice was definitely heard, and for the first time I was in discussions and roundtables that were looking at rare disease and the future of all policy in the USA moving forward. For the first time since our diagnosis, I felt I was steering the ship. I felt proactive instead of always feeling reactive to what new challenge Cystinosis presented us as a family.

CRN: What do you think events like this do for the cystinosis community?

Jay: They give perspective to our cause and reaffirm that rare diseases are actually not that rare. If the same research, determination, and push can be utilized in this arena the way we see public health policy affecting real change in cancer, HIV, and diabetes, etc. then the sky is literally the limit to realizing our dreams of eradicating rare diseases for the coming generations. In the end the Cystinosis community has shown through its research and grassroots initiatives that even a few can affect many. Treatments that have seen success in Cystinosis are at their core roots novel therapeutics for many other disorders that affect the lysosomal transport pathways. Truly, by opening up new funding avenues, allocating federal dollars towards establishing a rare disease center of excellence, and humbly teaching our elected officials how to truly speak for "all Americans" we can and will realize our not-so distinct dream of eradicating this insidious genetic disease.

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

By Jenni Sexstone, Treasurer

For the 3 months ended March 31, 2020

Revenues

Total income for the three months ending March 31, 2020, was $294,000 compared to $188,000 in 2019 due to increased grant payments as well as increased fundraising efforts.

Expenses

Total expenses of $49,000 were lower than expenses for the same period during 2019 of $112,000. Last year there was $70,000 of grant payments. All other expenses were $7,000 higher than the same reporting period in 2019 due to timing of newsletter expenses.

CRN had net operating income of $245,000 for the three months ending March 31, 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 March 31, 2020 was $430,000. Net change in cash for the first quarter 2020 was an increase of $235,000.

Research Update
2020 Call for Proposals

By Christy Greeley, Vice President of Research

Christy and Jack Greeley.

The Cystinosis Research Network 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.

CRN has funded over $4.5 million total in research grants and fellowships. CRN funded 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 is pleased to announce its 2020 Call for Research Proposals. Proposals should be submitted by August 14, 2020. Applicants must submit an electronic copy of each proposal to: Christy Greeley, Vice President for Research cgreeley@cystinosis.org.

The Scientific Review Board utilizes a two step-process to make funding recommendations. The first step occurs at the annual Scientific Review Board meeting. Proposals may be submitted to CRN at any time, however, CRN will issue a formal call for grant proposals prior to the annual meeting for preliminary review. The top rated applicants from the preliminary review meeting may be asked to present their proposals in person before the Scientific Review Board at its annual meeting, to be held in conjunction with the biennial conference. In years when the conference does not take place, the meeting will be held via conference call. After this second step, the Scientific Review Board will again rank the proposals and make its final funding recommendations to the CRN Executive Committee via the Vice President for Research.

The Executive Committee will meet within two weeks following the annual Scientific Review Board meeting

and will then vote on the SRB's recommendations. Funding decisions will be arrived at taking several factors into consideration. In addition to the SRB's recommendations, these factors may include such issues as the amount of money currently allotted for research funding, internal research topic prioritization, expressed wishes of donors, and efforts to support a wide variety of cystinosis researchers. More than one grant may be funded. A majority vote of the Executive Committee will be required in order to approve funding of any grant proposal. Decisions will be communicated to each applicant within two weeks following this meeting.

An optional, additional SRB meeting may be held at the discretion of the Executive Committee in the event that additional funds become available and outstanding qualified proposals remain unfunded. A call for new proposals may also be made at that time in the event that the amount of new funding available exceeds that required by any outstanding, qualified proposals. Funding recommendations would be arrived at after the optional meeting of the SRB. The Executive Committee would then utilize the same procedure in order to arrive at funding decisions.

PROPOSAL GUIDELINES

- Each application must include:
- Abstract/Summary of project
- Hypothesis and specific aims
- Methodology
- Data analysis plan and expected results
- Detailed budget — CRN will provide up to 10% of indirect costs
- Statement as to why Cystinosis Research Network funding is essential for the execution of this project
- Personnel (please include current CVs)
- IRB approval and consent forms, or plan to obtain such approval, when applicable
- Statement detailing other grant support available, if applicable
- Proposed start and end dates

CONDITIONS OF GRANT AWARD ACCEPTANCE

Upon acceptance of research grant funding by the Cystinosis Research Network, the awardee/recipient agrees to the following:

Progress reports will be required from awardees at specified time points. These will include reports at 6 month intervals (i.e. 6 month, 12 month, 18 month, etc.), for the duration of the study, as well as a final report at the conclusion. These progress reports should be an executive overview of the study's progress against key milestones, including budget and personnel updates. Budget payments will be linked to these reports.

The Cystinosis Research Network sponsors a Family Conference biennially. The Principal Investigator (or comparable level delegate) agrees to be available within reasonable limits of time and travel to present the findings of the research sponsored by the Cystinosis Research Network at this event. Reimbursement will be provided through separate funding for the Principal Investigator or delegate's expenses to attend.

Any publication of research funded by the Cystinosis Research Network must give proper reference to the Cystinosis Research Network. The Principal Investigator must mail a copy of such publications to the Vice President, Research and/or Executive Director of the Cystinosis Research Network.

REVIEW CRITERIA AND ASSIGNMENT OF PRIORITY SCORES FOR RESEARCH GRANT PROPOSALS

A single score is given to each individual proposal. This score is a numerical indicator of the best estimate of scientific merit. Reviewers use their own standards of excellence to arrive at this estimate of scientific quality and program cohesiveness using the set of adjectival descriptors below. Priority should be given to interventional research, both clinical and basic, that will lead to improved treatments for cystinosis. Only applications with significant and substantial scientific merit are scored. All other applications are "Not Recommended for Further Consideration".

Each reviewer votes on a scale of 1.0 to 5.0, using increments of 0.1.One (1.0) is the best score and 5.0 is the poorest among rated applications. The priority score of the Scientific Review Board is then calculated by averaging the individual priority scores and multiplying the result by 100.Ratings should be reached and recorded independently. Applications not recommended for further consideration are not rated.

Each reviewer is expected to use his or her personal standards of excellence in arriving at a priority score. Merit assessment includes such factors as how the recommended research would advance knowledge given the state-of- the-art in the disciplines involved; the likelihood of accomplishing this; the technical and scientific competence of the investigators; the degree of institutional support; the adequacy of facilities and resources; and the internal organization, administration, and quality control management.

The following shows the correspondence between numerical scores and adjectival descriptions:

1.0 – 1.5 Outstanding

1.6 – 2.0 Excellent

2.1 – 2.5 Very Good

2.6 – 3.5 Good

3.6 – 5.0 Acceptable

From the Fanconi Anemia Research Fund, Inc. guidelines, adapted from the peer review guidelines used by the National Institutes of Health, effective 1991; updated 5/96

Project Updates

Mechanisms Underlying Neurocognitive Changes in Cystinosis – Final Report

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

Departments of Neuroscience and Pediatrics, Albert Einstein College of Medicine, Montefiore Medical Center

Total Grant plus Travel Addendum: $338,322

With critical support from CRN, Drs. Foxe, Molholm, and Francisco from the Cognitive Neurophysiology Laboratory (CNL) in New York (https://www.cognitiveneurolab.com/; www.urmc.rochester.edu/labs/cognitive-neurophysiology.aspx) are characterizing perceptual and

cognitive function in cystinosis using cutting-edge non-invasive electrical brain imaging (scalp recorded electroencephalograms, EEG) and behavioral measures. In the past three years, they have tested a total of 42 children, adolescents, and adults with cystinosis from all over the country, plus an age matched comparison group of 50 individuals without cystinosis. This work has revealed novel information on how brain function is affected by cystinosis in children and adults. This work has been presented at the Cystinosis Family meetings in Utah and Philadelphia and several local and international scientific meetings, as well as published (or is being prepared for publication) in scientific journals. Here we present a summary of their findings to date.

Executive functions encompass brain processes that govern goal-directed behavior and serve to optimize performance on complex cognitive tasks, allowing one to behave flexibly and to adapt to novel, changing circumstances. Executive Functions are often affected in cystinosis. Executive function abilities—such as working memory, cognitive flexibility, and response inhibition—are critical for academic, professional, and social achievements. Response inhibition, the process by which one suppresses a well-learned response that might be irrelevant or inappropriate in a given context, is clearly essential for adjusting behavior dynamically with changing environmental contexts.

The CNL team has found that when it comes to response inhibition, there is a lot that works really well in cystinosis, in children and adults alike. Individuals with cystinosis can withhold their responses just as well as healthy controls. However, they make a few more errors, and brain activity shows that withholding a response takes more effort. Cognitive tests of executive function show that these individuals with cystinosis can perform the tasks just fine, but that it takes them a little bit longer. So, giving a little more time to execute tasks and process information may go a long way in academic success for children with cystinosis.

The ease with which we make sense of what we see and hear belies the complexity of auditory and visual processing in the brain. However, even basic recognition that the object you see in front of you is a cup of coffee involves a complex hierarchy of cortical brain processing that begins with extraction of the very simple features that comprise the object, and includes detecting where the event is, storing it in memory, comparing it with other events, and identifying it. Along the way you will likely even assign some value to it (caffeine = high value!). Brain processing can break down at any of these stages, and have cascading effects on later stages of information processing such as described under Executive Functions. The CNL therefore has also looked into auditory and visual processing in cystinosis.

For auditory processing, the CNL research team found that while basic auditory processing is fully functional, children and adolescents with cystinosis seem to have some difficulties with auditory sensory memory. Sensory memory is the shortest type of memory and it helps keeping track of sequences of information. Because basic and complex aspects of brain processing are related and co-dependent (for example, a problem in a more basic process can hinder the processing at a more complex level), difficulties at this very basic level of memory could impact, for example, working memory—crucial to fulfill day-to-day tasks and to academic success. Remarkably, though, in adults with cystinosis both auditory processing and auditory sensory memory appear fully intact. This means that though some difficulties are present in younger individuals with cystinosis, they appear to be resolved in adulthood. Since this could have a negative impact in both school and home contexts, this should

be considered when developing education plans.

Focusing in on visual processing, brain data from the CNL show that the visual response is substantially larger in individuals with cystinosis, and this is the case for both children and adults. This presents a powerful biomarker that can be used to see if treatments help to normalize brain function. This increased visual brain response may reflect reduced neural inhibition in visual cortex, which would lead to an imbalance of sensory processing in the brain. This in turn might be related to the impaired visuo-spatial ability that has been observed behaviorally in cystinosis. The team at CNL are conducting analyses of additional data to understand the basis of this difference, which may provide clues for treatments to counteract the impact of cystinosis on brain function.

In conclusion, we are pleased to report to you that the CNL teams' investigations into brain function are providing critical information on the strengths and weaknesses that are present, on average, in individuals with cystinosis. This information in turn hints at the types of additional cognitive support that will be of most use to affected individuals in our community.

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.

The next meeting of the CAB, originally scheduled to take place in July at the Cystinosis Network Europe International Conference, will instead take place virtually in June. 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 by Dr. Bill Gahl and Dr. Galina Nesterova. 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 an expanded Dialysis and Transplant section coming soon which will include a broad range of information and resources for those facing these challenges.

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 at https://www.cystinosis.org/support-resources/publications-guides/

Jack and Alex Greeley.

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 2.45 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**.

2019 Donor Honor Roll

**Network for Good represents all donations made through Facebook.*

$100,000+
Horizon Pharma USA, Inc.

$50,000-$100,000
Leadiant Biosciences, Inc.

Shared Health Alliance

$25,000-$49,999
Network for Good*

$10,000-$24,999
AVROBIO

Wyman Family Foundation

$5,000-$9,999

AllianceRx Walgreens Prime

Mylan Inc.

Recordati Rare Diseases Inc.

Schleuder, Don L.

The Precourt Foundation

$2,500-$4,999
Paypal Giving Fund

Roesler, Pamela and Jeffrey

Kaskel, Dr. Rick and Mrs.Phyllis

Carmichael, Scott and Tia

Gorkiewicz, Daniel & Christine

$1,000-$2,499
Riverside Gardens of Vero Beach

Russell, Jeffery M.

Raymond James Charitable Endowment Fund

Cash

Julian, Dan and Jan

Bank of America Employee Giving Campaign

LDC NWTF

Arndt, Frank and Raymond

Brandon Farris Inc.

Croce, Billy and Darlis Kay

Danzig, Robert

Exchange Club of Dearborn Foundation Inc.

Gilberg, Josh & Katy

Hughes, Carol Lee TTEE

Krieger, Scot W.

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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 (CRN) 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.