2021 Family Conference

This summer marks our 10th biennial conference. With the global pandemic casting doubt about the safety of in-person meetings, this will be our first time hosting entirely online. We’ve created a registration and attendee hub where you can access each opportunity available throughout the weekend. By registering, you’ll have access to the latest in research updates, breakout groups, cystinosis expert panels, and sessions tailored to multiple stages within the cystinosis journey. Anyone impacted by our rare disease is invited to attend.

For the conference agenda and information on how to obtain a computer and/or internet access at no cost, visit pages 3-7.
The President’s Letter

As much as I tried to start this letter without mentioning the pandemic – it just isn’t possible! How do you describe a year like we just experienced? Indescribable? Unique? Historic? Unprecedented? Like never seen before? From the pandemic caused by COVID-19; to one of the most highly contested presidential elections, to social injustices and unrest, to Zoom becoming a household name – the last 12-16 month period is one for the history books.

Without a doubt, COVID-19 has disrupted life as we know it. And while it has been a time of remarkable challenges, it was also full of resiliency in many respects. As much as we all want to attend an in person conference, when it became clear that it just wasn’t possible, your CRN Board pivoted and has planned a unique virtual conference for July. The family conference is an important ingredient to the CRN community and I hope that you will take the opportunity to connect with one another.

At its core, CRN is about community. Our family was fortunate to be welcomed by the CRN Community back in 2006. It seems like yesterday when Kacy was in the hospital as doctors attempted to figure out her medical puzzle. No doubt Kacy has been handed a difficult journey, but she isn’t alone. The CRN community has played an important role for our family, and I hope yours, by providing timely information, comfort, research, a sense of belonging and lifelong friends.

Kacy and others living with cystinosis deserve no less than our full commitment in achieving the CRN Mission: supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis. I have come to know and appreciate the dedication and hard work that the Board does on behalf of the cystinosis community. Since 1996, there have been countless volunteer hands pitching in to benefit our community. You
all deserve our THANKS! I would also like to recognize and thank current Board members for serving our community:

Jen Sexstone, Ina Gardener, Christy Greeley, Jen Wyman, Marybeth Krummenacker, Jonathan Dicks, Carol Hughes, John Maccarone, Gail Potts, Herberth Siegler, and Melanie Vachon.

We also owe a great deal of gratitude to our past and current medical advisors and scientific review boards. I invite you to visit the website (www.cystinosis.org) to learn more about our medical partners.

One of my favorite quotes is by Christopher Reeve: “Once you choose hope, anything is possible!” Working together I know that we can find improved treatments and accelerate the discovery to an ultimate cure. I should probably add... working together AND raising some funds. ☺ I recall standing in front of a few hundred people at one of our fun run/walk fundraisers and sharing that I’d like to say money doesn’t matter – but it does – it is what drives research for a cure. You all know that rare diseases simply do not receive the same attention or funding as other conditions. You can help too. Every dollar counts. You never know, it might be the front lawn lemonade stand that puts us over the top. If you are looking for fundraising ideas feel free to reach out to Board member Jonathan Dicks (Jonathan.Dicks@gmail.com).

Lastly, thank you for the opportunity to serve as Interim President. Being able to advocate for Kacy and others living with cystinosis is a privilege and honor. CRN is a special community and is making an impact. I hope to “see” you all in the Zoom hallways during the virtual conference (register at https://cvent.me/2KNqR0). The Beat Goes On!

Proud to Serve,

Tim Wyman

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Cystinosis Research Network 2021
Virtual Family Conference Agenda

**Friday, July 16th**

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tbody>
<tr>
<td>6:00 pm – 8:00 pm</td>
<td>Conference Kickoff/Family Introductions</td>
</tr>
<tr>
<td></td>
<td><strong>Hosts:</strong> Jonathan Dicks, VP Development and Jen Wyman, VP Family Support</td>
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<tr>
<td></td>
<td>Please join us for welcome and overview of the conference and family introductions. Bring your entire family!</td>
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**Saturday, July 17th**

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
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<tbody>
<tr>
<td>10:00 – 10:30 am</td>
<td>Welcome and CRN Overview</td>
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<td></td>
<td><strong>Tim Wyman, President</strong></td>
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<tr>
<td>10:30 am – 11:00 am</td>
<td>Cystinosis – A Review of Old and New</td>
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<td><strong>J.J. Zaritsky, MD, PhD</strong></td>
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<td></td>
<td>A review of the epidemiology, pathophysiology and treatment options of Cystinosis.</td>
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<tr>
<td>11:00 am – 11:30 am</td>
<td>Anticipating Renal Replacement Therapy/Stanford protocol combined stem cell transplant and kidney transplant from same living donor</td>
</tr>
<tr>
<td></td>
<td><strong>Paul Grimm, MD</strong></td>
</tr>
</tbody>
</table>
11:30 am – 12:30 pm

Panel Session 1

Panel Sessions are targeted towards different stages of the Cystinosis journey, divided into tracks by age. They will be smaller group sessions with a panel of experts presenting information, guiding discussion and fielding questions. Attendees should feel free to attend sessions of most interest to them.

Caregiver/Infant & Child (0-10): Cystinosis 101
Moderator: Jen Wyman, VP Family Support
Panelists: J.J. Zaritsky, MD, PhD Craig Langman, MD, Ranjan Dohil, MD Paul Goodyer, MD, Ewa Elenberg, MD, MEd, Rachel Bishop, MD

Teenager (11-17): Cystinosis 201
Moderator: Herberth Sigler, Board Member
Panelists: Rick Kaskel, MD, PhD, Larry Greenbaum, MD, PhD, Mihir Thacker, MD, Minnie Sarwal, MD, FRCP, DCH, PhD

12:30 pm – 1:00 pm

Break

1:00 pm – 2:00 pm

Panel Session 2

Panel Sessions are targeted towards different stages of the Cystinosis journey, divided into tracks by age. They will be smaller group sessions with a panel of experts presenting information, guiding discussion and fielding questions. Attendees should feel free to attend sessions of most interest to them. Some sessions are closed as noted below.

Caregiver/Infant & Child (0-10): Neurocognitive & Educational Issues
Moderator: Jonathan Dicks, VP Development
Panelists: Doris Trauner, MD

Teenager (11-17): Transplant and Dialysis
Moderator: Jenni Sextone, Board Member
Panelists: Paul Grimm, MD, Rick Kaskel, MD, PhD, Roz Mannon, MD, Ewa Elenberg, MD, MEd Minnie Sarwal, MD, FRCP, DCH, PhD, Larry Greenbaum, MD, PhD

Adult (18+): Living with Cystinosis as an Adult: Healthcare, Expectations, Self-Management, Reproductive Issues, Mental Health Issues
(closed session for cystinosis adults and their partners)
Moderator: Sara Healy, Adult Leadership Advisory Board (ALAB) Member
Panelists: Maya Doyle, MSW, PhD, LCSW-R, Bill Gahl, MD, PhD, Galina Nesterova, MD, FABMG, Elena Levetchenko, MD, PhD
1:00 pm – 2:00 pm, continued

Parents of Adults with Cystinosis
(closed session for parents of adults with cystinosis)

Moderator: Carol Hughes, Board Member

The purpose of the session is intended to be a private setting for parents of adults with cystinosis to share information, insight, advice and encouragement. This session will provide an opportunity to learn from others who face similar challenges, and allow you to talk about your experiences.

2:00 pm – 3:00 pm

Medical Panel

Moderator: Bill Gahl, MD, PhD

Panelists: Paul Grimm, MD, Craig Langman, MD, Jess Thoene, MD, Rick Kaskel, MD, PhD, Rachel Bishop, MD, Doris Trauner, MD, Larry Greenbaum, MD, PhD, Paul Goodyer, MD

Please join for the unique and informative opportunity to have your questions and concerns addressed by the leading physicians and researchers in Cystinosis. Questions will be gathered during the course of the conference utilizing the chat function.

Saturday, July 17th

(All times Eastern Daylight Time)

10:00 am – 10:05 am

Welcome

Christy Greeley, VP Research, Executive Director

10:05 am – 10:20 am

Cystadrops Update

Recordati/Anovo

10:20 am – 10:35 am

Cystaran Update

Leadiant

10:35 am – 10:50 am

Procysbi Update

Horizon Therapeutics

PAL program, update on WBC kits

10:50 am – 11:20 am

Stem Cell Gene Therapy for Cystinosis

AVROBIO

11:20 am – 11:50 am

Disease Specific Quality of Life Questionnaires

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis

Newborn Screening Update

Katharina Hohenfellner, MD, Harald Schubert, Novotec

Cystinosis Research Network Funded Investigator

11:50 am – 12:20 pm

Cognitive Control Systems in Cystinosis

Sophie Molholm, PhD, John Foxe, PhD

Cystinosis Research Network Funded Investigator
12:20 pm – 1:00 pm  Break

1:00 pm – 1:30 pm  Validation Study of a New Biomarker for Cystinosis in a Retrospective Cohort from NIH

Elena Levchenko, MD, PhD
Cystinosis Research Network Funded Investigator

Measuring while blood cell (WBC) cystine levels is a golden standard for the diagnosis of cystinosis and for monitoring cysteamine therapy. However, the laboratory technique for WBC cystine determination is laborious and is only available in a limited number of specialized laboratories. Moreover, because the pool of WBC cells is frequently renewed, WBC cystine levels reflect a short period of (adequate) cysteamine therapy. Our group has searched for alternative biomarkers of cystinosis and has found that the enzyme chiotriosidase (chito) might be a suitable candidate. Chito is released by activated macrophages which try to clear cystinosis tissues from cystine crystals. From the technical point of view, chito has several advantages such as the availability of easy and cheap method for its measurement, and the fact that the enzyme is extremely stable, allowing sending blood across borders on dry blood spots. We have found that plasma chito activity correlate with WBC cystine levels and kidney function of cystinosis patients in a cross-sectional (Elmonem et al. 2016) and a longitudinal (Vest et al. 2020) cohorts. Additionally, the occurrence of the extra-renal complications was associated with higher chito levels in adult patients (Vest et al. 2020). In the current project supported by CRN we aim to validate our findings in a large retrospective cohort of cystinosis patients followed at the NIH during a very long period of time. This long follow-up study aims to validate the clinical utility of chito as a novel long-term biomarker of cystinosis.

Project title: Chitotriosidase as a therapeutic monitor for cysteamine therapy in cystinosis: a retrospective.

1:30 pm – 2:00 pm  Perturbations in the V-ATPase Pathway Drive Pathology in the Male Reproductive System in Cystinosis

Minnie Sarwal, MD, FRCP, DCH, PhD
Cystinosis Research Network and Cystinosis Ireland Funded Investigator

2:00 pm – 2:45 pm  Parents of Children and Adults with Cystinosis Panel

Moderator: Jen Wyman, CRN VP Family Support
Panelists: Kristina Sevel, Jill Morrill, Chelsea and Brian Meschke

Panel presentation during which parents of children and adults with cystinosis will answer prepared questions and address topics related to the use of coping mechanisms through the ups and downs that cystinosis brings related to not only developmental and transitional issues of daily life but also medical issues. Parents of individuals at every stage of the disease will be featured and will share how they have managed the variety of challenges they have faced.

2:45 pm – 3:30 pm  Adults Living with Cystinosis Panel

Moderator: Maya Doyle, MSW, PhD, LCSW-R
Panelists: Hannah Creel, Brianna Dundon, Christina Morris, Brian Ensor

Panel presentation during which individuals living with cystinosis who serve on CRN’s Adult Leadership Advisory Board (ALAB) will answer prepared questions and address topics related to the use of coping mechanisms and strategies for success through the ups and downs that cystinosis brings.
Started in 2018, Cystinosis Awareness Day was created to educate the general public and medical communities about cystinosis. May 7 (5/7) was chosen to represent the most commonly linked cystinosis mutation, the 57-kb deletion.

In four short years, this day has transformed into a highly anticipated event. We've received support from national groups like the American Association of Kidney Patients and American Kidney Fund and have reached over 20 countries with cystinosis patients embracing this day of recognition.

The theme for 2021 was myth-busting. Caregivers, those living with cystinosis, and healthcare professionals answered the call. Most awareness work happened online by sharing misconceptions about cystinosis through our website, blog posts, emails, TikTok videos, Facebook shares, retweets, and Instagram stories. This multi-channel approach resulted in thousands of views and impressions.

To honor the fourth year, the CRN secured donors ready to match contributions, ensuring each dollar would go four times as far to support vital community programming and research. Projections currently have us exceeding $23,000 in donations.

We invite you to view the Cystinosis Awareness Day myth-busting videos from Dr. Bishop, Dr. Elenberg, Dr. Trauner, the Albert Einstein School of Medicine Team, Dr. Thoene, and Dr. Maya Doyle on our YouTube channel at http://bit.ly/2XG8fbk.

Thank you cystinosis warriors and those who continue with us through the fight, May 7th and every day!
Succeeding with Cystinosis: ALAB Commemorates Cystinosis Awareness Day

The Adult Leadership Advisory Board (ALAB), our umbrella group with a membership made entirely of adults living with cystinosis, re-launched their “Succeeding with Cystinosis” campaign for Cystinosis Awareness Day. They challenged the community to search for the successes and failures experienced over the past year. Learn more by visiting cystinosis.org/alab or the Adult Leadership Advisory Board page on Facebook.

Interested in becoming a part of Cystinosis Awareness Day 2022? Email info@cystinosis.org.
Cystinosis Awareness Day in Mexico

By Victor Gomez

As part of “Cystinosis Awareness Day” May 7th, 2021 a round table discussion was held by cystinosis experts:

• Dr. Alfonso Huante Anaya, Pediatric Nephrologist National Institutes of Pediatrics
• Dr. Mara Medeiros Domingo, Research Unit, Mineral Bone Metabolism, Pediatric Nephrologist, Children Hospital Mexico
• Dr. Gloria Beatriz Hernandez, Silverio Chief Pediatric Unit, Pediatric Nephrologist, Juarez Hospital Mexico
• Dr. Erika Vargas Quevedo, Pediatrician, Research in Lisosomal Storage Diseases, Coordinator in Inborn Errors Metabolism and Rare Diseases Clinic, 20 Noviembre Hospital, Mexico

They conducted a round table to talk about nephropatic cystinosis:

What is cystinosis?, Diagnosis: Clinical, Biochemical and Molecular, Treatment of Fanconi Syndrome and Indicated treatment for Nephropatic Cystinosis.

During those two hours we had the opportunity to listen to medical experts in the field on the topics mentioned above. Attendees had the opportunity to have a question and answer session, which were answered in a timely manner and there were issues that remained on the table which will be discussed very soon.

We thank the speakers for their willingness and support in the realization of this first cystinosis webinar, as well as all the attendees, doctors, students, patients and families. We are very pleased that, in our country, the number of physicians helping patients manage cystinosis is increasing. The main objective of our organization is to carry out this type of events is to raise awareness and inform about this rare disease.

Complimentary Care Package Program

The Care Package Program was created to help individuals and families with cystinosis during difficult times. We will send a personalized package with educational materials and supportive tools to you -or- a loved one. If you or someone you know has recently received a diagnosis, is actively listed or awaiting transplant, or is suffering the recent loss of a loved one with cystinosis, request your box at cystinosis.org/care-package.
On October 15th, 2019 the world lost a soul that had an amazing love of life. Laura Kathryn McGinnis enjoyed being alive more than most people I know. The last phone call we had, before the amoeba that would take her life destroyed her brain, she thought she was in New York City with her friends. Why was I bugging her when she was having fun? This is something that I hold onto. That in her last coherent thoughts and spoken words she was living fully, if only in her mind. Adventure and grabbing every experience she possibly could have was what drove her and on the days when the hole she left feels too big to bear I imagine she is on her greatest adventure. As her beloved Albus Dumbledore said, “To the well-organized mind, death is but the next great adventure”.

To honor the life she so fully lived, my hope was to capture some of her by helping others pursue their adventure. The Live Like Laura Fun Fund (LLFFF) goal is to enable the person with cystinosis or the sibling of someone with cystinosis to reach for all that life has to offer, to have fun, and to make memories. In her short life funds and yes, sometimes her health, were the only factors that ever stopped her so to help others reach for their adventure became my goal to honor her memory. An applicant can request up to $1,000 USD every 3 years. So far, the LLLFF has been honored to help three individuals pursue their interests and to travel. One person will be renting a camper van to explore their region, one person will be starting a podcast, and one person will be taking horseback riding lessons. A fourth, soon to be awarded recipient, will be visiting family in another country to mourn together the loss of someone they loved.

Each application is assessed by a committee of Laura’s friends and family and each year we will award as many as we can based on the funds available. As you can tell by the recipients to date, the request is what is important to the individual, their dream, desire, or hope. The LLLFF hope is to be a “yes” in a world that sometimes feels full of “no’s”. We hope to share photos of awardees in the future and their words on the adventure that was attained; with each story I know my girl will be grinning. Every day we are here is a gift, an opportunity to be our best selves. I hope that we can all learn from my precious squirrel, to live fully, to love without boundaries, and to say yes to every adventure possible.

Life is short, I hope you live it. Submit your application online at cystinosis.org/lcff.
Cystinosis Network Europe (CNE) Update

By Denise Dunn

Cystinosis Network Europe (CNE) has continued to work with the healthcare industry through the Community Advisory Board (CAB) to discuss the issues of most importance to our community. In April and May the CAB members (14 representatives from seven countries) met with two pharmaceutical companies to discuss their plans for engaging with the cystinosis community and potential access to therapies. The CAB raised many issues of concern to those living with cystinosis which we hope will ultimately make better therapies more quickly available.

Cystinosis Network Europe International Conference 2022

We are delighted to announce that the 2022 Cystinosis Network Europe (CNE) International Conference will take place on 7-10 July, in Leuven Belgium, hosted by our colleagues in Cystinose Groep Nederland en Vlaanderen. We look forward to our friends and colleagues joining us for the scientific meeting on 7 July and the family conference from 8-10 July. Covid protocols will be carefully monitored and adhered to and we will update the CNE website (www.cystinosis-europe.eu) as details for the event are confirmed.

Dublin Cystinosis Workshop 2021

By Denise Dunn

The 7th Annual Dublin Cystinosis Workshop (DCW) went virtual again in 2021. On 20th May, Cystinosis Ireland hosted the biggest cystinosis research event in Europe this year, with 66 researchers, clinicians, and patient experts from 14 countries joining on Zoom. The first of the keynote speakers, Prof. David Sabatini of the Whitehead Institute, MIT, was a new face to many of in the cystinosis world. His lab specialises in mechanisms that regulate physiological and pathological growth and metabolism in mammals, with an interest in the signalling pathway anchored by the mTOR kinase, which David discovered when he was an MD/PhD student with Dr. Solomon Snyder at Johns Hopkins Medical School. David’s keynote address focused on the MFSD12 mediation of cysteine importation.

The second keynote speaker was one more familiar to many in our community. Prof. John Foxe of the University of Rochester School of Medicine and Dentistry discussed “Exploring the Neurophysiology of Cystinosis.” Prof. Foxe was joined by his colleague Dr. Ana Francisco to answer questions and discuss some of the issues raised by their research.

The DCW continues to be a valuable forum for researchers new to the field to introduce their ideas and gain feedback from their peers on the work they are undertaking. Our well-established colleagues continue
to share their knowledge and expertise generously. en speakers presented short communiques of the new research results and novel and exploratory research directions they are undertaking, with Dr. Jennifer Hollywood of the University of Auckland returning as last year’s winner of the Prof. Roz Anderson Memorial Prize. The presenters of these short communiques were eligible for the 2021 prize and this year Tjessa Bondue, a new researcher in the area of cystinosis, was the winner. You can see more about Tjessa’s work here - https://bit.ly/2S4iTXH.

Two other sessions rounded out the day. The first was a discussion of the potential to create a cystinosis clinical trials hub in Ireland. Presenters from the National Office for Research Ethics Committees and the Health Products Regulatory Authority, both state bodies instrumental in the development of a suitable environment for clinical research in Ireland, explored and explained how clinical research infrastructure works and can support research here. Prof. Francesco Emma also joined the meeting to share the importance of a worldwide patient registry and how that can and should assist trials.

The final session was updates from clinical trials being planned or currently under way. Prof. Herbie Newell (University of Sunderland) discussed his team’s work on the pro-drug CF-10; Prof. Paul Goodyer (McGill University) updated on the development of a drug therapy for the nonsense gene mutation; Prof. Anuj Chauhan (Colorado School of Mines) discussed his plans for cystinosis therapy by contact lenses; Prof. Stephanie Cherqui (UCSD) discussed the hematopoietic stem cell gene therapy for cystinosis with some updates from the phase I/II clinical trial; and Cecile Berends updated on a novel sustained release cysteamine bitartrate formulation for the treatment of cystinosis. It was extremely exciting and interesting to hear of the work being done across the world aiming to bring new and more tolerable treatments to people living with cystinosis.

Cystinosis Ireland would like to thank most sincerely the excellent meeting chairs, Prof. Atif Awan (Children’s Hospital Ireland, Temple Street) and Prof. Elena Levchenko (KU Leuven, Belgium) who were ably assisted by Dr. Patrick Harrison (University College Cork) and Dr. Jennifer Hollywood (University of Auckland), for all their hard work, and in particular keeping the meeting on time! The conference committee also included Dr. Thomas McDonald (Bristol Myers Squibb & Cystinosis Ireland board member); Dr. Achim Treumann (KBI Biopharma BV & Cystinosis Ireland scientific advisor); Anne Marie O’Dowd (Cystinosis Ireland board and scientific committee member, Chair of Cystinosis Network Europe); Dr. Ruth Davis (Cystinosis Ireland research manager) and Denise Dunne (Cystinosis Ireland operations manager).

Pediatric Academic Societies 2021

By Carol Hughes

Keeping in stride with all the other anomalies this past year, the Pediatric Academic Societies (PAS) annual conference was held as a virtual event.

The PAS connects thousands of pediatricians and other health care providers worldwide. This international gathering offers opportunities for a global audience of physician-scientists, clinicians, and educators to share research, explore new ideas, build career opportunities, and collaborate on future projects. Presentations cover issues of interest to generalists as well as topics critical to a wide array of specialty and sub-specialty areas.

CRN has been fortunate to participate exhibiting in these annual symposiums for nearly 20 years. This has given us an excellent opportunity to personally meet with attendees on an individual basis forging meaningful relationships with physicians and other professional attendees sharing the latest information on cystinosis research along with literature for them to share with their patients and colleagues.

This year was by far quite different than all other meetings. We were able to have a ‘booth’ same as prior years, but it we truly missed the valuable in-person interaction with those that ‘stopped’ by our exhibit and being able to physically share our literature and other giveaways. Thankfully the literature is available in our ‘booth’ and also at www.cystinosis.org. Gail Potts, Heidi Hughes and myself ‘manned’ the CRN booth during four days of the Virtual Exhibit Hall the end of April through May 4th. Looking forward to next spring when PAS 2022 meets in Denver, CO.
As I begin this article I am so very honored to be a part of an organization that continues to offer so many opportunities to our community. Whether reviewing the applications for scholarships, planning our first ever virtual conference, awarding families over 30 PCs... all extraordinary opportunities for the cystinosis community!

We also continue to represent the rare disease community in a variety of ways.

• **Inclusivity in Patient Advocacy**

Horizon sponsored virtual meeting – March 15. In light of COVID-19, it was a very enlightening discussion about inclusion for all under-represented groups. Finding ways to include some of these groups and making sure they are participating not only when it comes to COVID-19 and vaccine availability, but overall keeping people engaged in these ongoing discussions.

• **Every Life Foundation/Genetic Alliance sponsored virtual meeting**

“Rare on the Road” – March 23. An ongoing discussion of training the next generation of advocacy leaders. This included basic information from speakers on how to tell your rare disease story and the impact it can have, maintaining the relationships with groups and encourage involvement in their own rare disease space but the “umbrella” space as well.

• **National Health Council**

The CRN was approved to join as a member of the National Health Council. I personally spoke with their Associate Director of Membership about our application back in January.

He strongly encouraged us to submit it and we did. We were recently approved at their last membership meeting. The NHC has been around since 1920 and is a highly respected policy organization in Washington, D.C. They have a high level of standards that is expected of membership and CRN met them as far as accountability to our community and transparency. They have an online symposium coming up in the fall, “The 2021 Science of Patient Engagement Symposium – Achieving Trust in and Trustworthiness of Science.” With the overall skepticism about the COVID-19 vaccinations, this will be geared towards trusting and respecting the science and the outcomes of medical research.

CRN continues to maintain relationships with various organizations: NORD, Genetic Alliance, Every Life Foundation and personal contacts within the rare disease community.
Family Support Committee Update
By Jen Wyman, Vice President of Family Support

It has been a trying year to say the least. Stress can and has gotten the best of us. It is in those times where we have to remember we are all in this together. We are a very fortunate group of people, sharing our very unfortunate journey with cystinosis, together. We are literally just a click away from finding someone who understands and supports us. We are a family, brought together by a common thread. We are a melting pot of people... different, yet the same.

I discovered a blog a few years back by Mark and Angel Hack (marcandangel.com). They are full of wisdom and daily inspiration and somehow have delivered messages at just the right times I needed them. One of the most recent was this…

"When life is pressing you and stressing you out, pause, take a deep breath, and remind yourself that you are not at the center of the universe. When you’re overwhelmed by life’s daily struggles, it’s so easy to feel like you’re at the center. But you aren’t. None of us are. Truth be told, we all have the occasional tendency to put ourselves at the center and see everything in life from the viewpoint of how it affects us. This can have all kinds of unfavorable effects, from feeling sorry for ourselves when things don’t go exactly as planned, to doubting ourselves when we fail to be perfect. Finding little ways to help others snaps me out of my self-centered thinking, I start to think about what others need. The central question now becomes - How can I give back?"

Our daughter, Kacy, is graduating from high school this year and will head to Grand Valley University in the fall to pursue a degree in nursing. It hardly seems possible and we couldn’t be more proud of the person she has become. She has embraced her life with cystinosis. It isn’t who she is, but rather a part of who she is. Cystinosis has molded her, guided her and challenged her and while it hasn’t been smooth and easy it has made her strong and independent and resilient and wise.

I have had conversations with many of you who are embarking on this journey. It is NOT an easy one. It is NOT for the weak of heart. It is NOT fun. Watching your child struggle and suffer is a parent’s worst nightmare. But it does get better, more manageable, and it gets easier to breathe. And you are NOT alone. When you discovered the Cystinosis Research Network you opened up your world to a new family-
one that will support you and guide you. It is a family with a strong will, big shoulders and listening ears and huge hearts. You will hear over and over again... “You got this”. And you do.

Our family is entering a new chapter. As parents we loosen our reigns a bit. We hope and pray that we instilled the lessons she needs to move forward, apart from us. Tim and I have leaned on many of you who have done this before us... lifted us up in our darkest moments and celebrated with us in moments of victory. We “need” our cystinosis family in a different capacity now. Kacy needs it in her own way and will find her place within it. It is the reason we stay on the Board and stay active in the community. We are better people to have been supported and guided by this family and it is our turn to give back to those of you who need it now.

We welcome, love and support the new families and we mourn with those who have lost loved ones. It is a battle we would never want to fight alone and we are grateful for all of you.

Support While You Shop with Amazon Smile

Did you know? A percentage of your Amazon purchases could help support our research and programming efforts - at no additional cost. Here’s how:

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

Happy Shopping!
Our Introduction to the World of Cystinosis

My name is Victoria Westfall. I am a 23 year-old single mother to a beautiful little girl named Addyson and we are from a small town in the Ohio Valley. She turned two years-old in March but our search for a diagnosis for Addyson started not long after I brought her home after birth. First, she started to vomit relentlessly, and this would continue for her entire first year of life. At two months old when I brought my concerns to a doctor, they diagnosed her with acid reflux after a hospital stay and insisted she would grow out of it. However, she continued to show signs of illness and did not simply "grow out of it", causing me to switch pediatricians to find the answers I was looking for. At Addyson's 15 month check up, I noticed she had failed to grow or gain weight in comparison with her 12 month check up. This is the point where her father and I started to become even more concerned about the health of our daughter.

Finally, after no answers, we switched pediatrics for a second time and found again at her 18 month checkup that she had hardly grown in height or weight in eight months time. After this, and a few months of testing and talking with our new pediatrician, he referred us to a Children’s hospital endocrinologist. There, the endocrinologist decided to refer Addyson to a nephrologist. After lots of testing, on May 3, 2021, Addyson was diagnosed with Fanconi syndrome, Type 2 RTA, and bone rickets. On May 6, 2021, I graduated college for Medical Coding and Billing. On May 7, 2021, Addyson was hospitalized. During this hospitalization on May 10, 2021, she was finally diagnosed with cystinosis. On May 12th she had surgery to have a g-tube placed for medication administration and she was discharged to go home on May 14th.

She is currently on several medicines and we are fighting our insurance company to get her Procysbi started, a very important drug in the treatment and maintenance of her disease. Addyson is an amazingly smart and resilient little girl, and has adapted to her g-tube and new way of life with ease. I am amazed by her every single day. I myself have been very overwhelmed (to say the least), during this entire process that I’ve been fighting for my little girl. However, every day does get easier. Before Addyson’s May 10th diagnosis, I had never even heard of cystinosis. Now every day since, I strive to learn something new about this disease to advocate for my daughter and those like her. I want to do everything I can to raise awareness for my baby and the babies of all the other mothers and fathers who have sat up at night with their sick child praying for an answer. Every single night I pray for the day there is a cure available for everyone affected by cystinosis.
Our family’s journey with cystinosis began in January 2021, when Briley’s pediatrician discovered she had lost a pound at her 18-month well child check. Other than frequent thirst and urination, we had no notable cause for concern regarding Briley’s health or development. With an abundance of caution, her pediatrician ordered labs that surprisingly pointed us to low bicarbonate and RTA. From there, we met with nephrology and through many more labs, determined Fanconi syndrome and began the exhausting rule out process to identify the underlying cause.

From the potential diagnoses on this list, cystinosis was the one we were most afraid of. We celebrated when ophthalmology told us she had no corneal crystals, but soon after we were crushed to learn that her WBC cysteine was elevated, though mildly; a recheck revealed an even lower number (both labs sent to Baylor). And so began the rollercoaster- we (and our nephrology team) were scratching our heads knowing that there would be no other reason for cysteine to be elevated, though her lack of other symptoms didn’t fully align with cystinosis. In early May, after 5 grueling weeks of awaiting genetic testing results, we confirmed her diagnosis of cystinosis, along with a more appropriate WBC cysteine result from UCSD.

Despite copious amounts of bicarb replacement, our girl continues to be vivacious- at almost two years-old her personality has really started to shine, and we have fun! We will soon begin Procysbi and look forward to leveling out this bumpy road.

Connecting with other families in the cystinosis community has been a game changer from Day 1 - we would be totally lost without this group! We are also incredibly optimistic about the developments currently underway in managing cystinosis, and are hopeful that the cure is on the horizon for Briley and all others affected by this disease.

“We have accepted the challenge of incorporating this disease into our lives, rather than incorporating our lives into this disease.”
Rare Disease Day - February 28, 2021

The goal of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients’ lives. This year’s Rare Disease Day theme was: Rare is many. Rare is strong. Rare is proud. We were certainly proud to see a young man with cystinosis featured by EURORDIS as part of this year’s event. Here is his story from rarediseaseday.org.

I AM REZA

My name is Reza, I am a 13-year-old living with cystinosis, in Iran with my family. I love going to the cinema, watching movies and reading books. I am also very fond of singing and dancing and really enjoy happy music. I really want to become an actor! I like taking part in acting classes, I think I am ready to star in a movie.

Each year I join Cystinosis Symposium which helps me meet other people living with a rare disease and I make lots of new friends there. We talk to each other and share our stories.

Generally, I really enjoy going out and attending social events. However, my recent kidney transplant and the COVID-19 pandemic have restricted my ability to socialise with all my friends at the moment because I have to be extra careful. Sometimes joining art, music and other educational classes can be difficult. I enjoy my friends and others don’t pity me, they consider me a strong boy.

My parents and family members have my back. I appreciate the Rare Diseases Foundation of Iran (RADOIR), the Iranian Cystinosis community, my doctors, especially the ones who did my kidney transplant, and all the people who have helped me. I will try my best to be a successful person in the future and make those around me happy.

- Reza, Iran
Rare Disease Day - February 28, 2021

RARE IN 60

Leading up to Rare Disease Day, we invited everyone impacted by rare disease to share their “Rare In 60.” This could be in the form of a 60-second video, 60-second photo collage or 60-word summary of what you’d like others to know about cystinosis/the rare disease that has impacted your life.

Paula Shal, Andrea Carr, Mason Stilke, Jessy Magnus, Laura Krummenacker, Jenn Loglisci, Hannah Creel, and Dr. Nesterova’s daughter shared their thoughts through song, video, and quotes. We published the contributions from Cystinosis Ireland and our rare disease colleagues Taylor Kane and Seth Rotberg from Remember the Girls and Our Odyssey (respectively).

The Adult Leadership Advisory Board (ALAB) launched a fundraiser in honor of Rare Disease Day. Every $5 donation received earned an entry to win a $100 Amazon gift card and some CRN swag. Congrats to the winner, Christine Sheppard!

We appreciate all who participated in making Rare Disease Day a success!

Feature Your Story Here!

We’d love to hear from you. We are always looking for the latest news from around the global cystinosis community to share with our readers… this includes your story! Send your ideas to info@cystinosis.org
I was trying to think of a way to honor my daughter Laura’s 35th birthday this year and to celebrate her life in a way to appreciate the fact that she is here…despite the odds that I was told when she was diagnosed in 1989 at the age of 3 ½.

I have been thinking a lot about Rare Disease Day at the end of February and coincidentally Laura’s birthday is February 13… right there in the middle, so I am realizing what a lucky month February is in my life! I was reminded once again of a piece of legislation that had a direct impact on the cystinosis community…the Rare Disease Act of 1983, and after recently listening to Abbey Meyers (NORD founder) on a recent webinar simply tell her story of “just being a grandmother from Connecticut”, I think of how lucky our tiny little rare disease community has been and for me personally, extraordinarily lucky!

I am always inspired when I hear Abbey tell the story about how the Orphan Drug Act of 1983 came to be. For anyone old enough to remember or young enough to Google, look for an old TV show called Quincy, M.E. Jack Klugman was the star and he wrote two episodes with his brother Maurice Klugman, who happened to have a rare cancer. That ONE television show was the pivotal point of pushing the Orphan Drug Act of 1983 through Congress. It is probably one of the best stories about the power of ONE and how it has had a direct correlation to having changed the lives of thousands of people living today with rare diseases still and how it played a major part in lives of the cystinosis community and still does today after 38 years! A startling number that is glaring to me is the fact that in 1983 there were just 10 new products approved by the FDA to help those living with rare diseases. Today that number is nearly 1000! While that doesn’t seem like enough and many of the rare disease community can agree things should be faster, science and research do seem to work at a snail’s pace! But science and research IS a process and safety and efficacy are the paramount driving forces with any new product before approval.

Hence, the reason it took 20 years to get approval on the Cystaran eye drops! But it was the long standing commitment between the drug company and the patients and doctors at the NIH that this happened and I am eternally grateful they did! Considering that there are over 7,000 rare diseases affecting over 30 million people, it is a miracle that the cystinosis community has the number of FDA approved treatments at all; Cystagon, Cystaran, Procysbi and now Cystadrops. We are a worldwide community of approximately 2,000 people with approximately 600 in the USA! I know this is a lot of numbers but we as a community are blessed and I believe that to be true, but if it wasn’t for that ONE piece of legislation from 1983, we probably would have NOTHING. As a historical FACT, Cystagon is #41 on the list of the first 100 FDA approvals after that legislation became law! To me, that is a miracle!

I am truly blessed that we will be celebrating Laura’s 35th birthday on February 13th! Without the brilliant minds of the many doctors in her life, the medications she takes and most importantly, without the research that was on-going and still is, without the critical partnership between advocacy partners and pharmaceutical industry leaders, we would have NOTHING and I know she would not be with me today! Laura is a survivor; rare disease, kidney transplant, breast cancer survivor and still going strong with a smile on her face and an attitude that tells me she will not be a victim of her circumstance! Laura is an incredibly strong person who I get my strength from every single day and we both consider every day to be a gift and a blessing.

Wishing a very Happy 35th Birthday to the ONE and only Laura Elizabeth Krummenacker!
Meet Bentley! He is the light of my life and my biggest hero!

He was diagnosed with cystinosis in October 2014 when he was two and a half, but was actually diagnosed with fanconi syndrome and rickets disease first in February 2014 when he was 18 months. They could not diagnose him with just a cystine level test, his level was 0.57, so we had to wait for an appointment with the genetics team. We spent a month in the hospital in February, with 3 surgeries during that time. He had a g-tube placed as well as a mediport. Those were the best decisions we could have ever made for him to help him through the tough start.

He will be 9 years old on July 10th, and he is doing extremely well. He has been on growth hormone for a year and a half and has grown over four inches in that time, the most he has ever grown so fast. He has an awesome in home nurse come once a month to our house for his labs to be drawn. We are eagerly waiting for one of the trials to become available for him. He is the rare of the rare, he doesn't have the typical 57kb deletion like most, he has a nonsense and a missence mutation.

Our biggest struggle with him lately has been with him asking questions like, “Mom - is cystinosis going to kill me?” or “Why did god make me like this and none of you?” It’s extremely hard to answer questions like this to an eight year-old little boy who all he wants to do is grow up, have kids, be in the military or become a police officer. But even with everything that he has been through, having good days, and having the terrible days cystinosis brings to him, he wakes up with a smile on his face every morning. He is so happy and loving all the time. He has a heart of gold and a personality everyone needs in their lives!

This journey has not been easy for him, or any of us, but it has definitely taught us to love like there is no tomorrow.
Diagnoses: Dealing with Cystinosis and Dyslexia

By Jessica Magnus

This article is to share our experience when cystinosis finds and crosses paths with Dyslexia which makes the whole situation much harder than the challenges of our children receiving their medicines during class time.

We all know that following the treatment of cystinosis requires a lot of physical and mental effort and having to focus on another learning problem is too much, for the person suffering from this disease and for the parents.

Since our children entered school, we knew that they could qualify for an IEP just because they have cystinosis, we are convinced that the school and the team in charge of providing your child with all the therapies that they need, would do a great job in providing all of this inside the school. But, what happen if this safety net fails?

Something like this happened in our case with Martina, since she entered school, she had an IEP where she received the therapies she needed during that time, I was very grateful since she was given for almost three years the opportunity to have a personal nurse with her at all times in the school, it was a great feeling for me to know that there would be someone with her all the time, plus that she would receive both OT and PT.

As time passed, Martina suffered a lot when she was doing homework, since it was quite difficult for her to write and read correctly, in addition, mathematics was not something that she did easily, even though she was already in a grade which she was supposed to write and read well for her age. Every afternoon we sat down to do her homework together, help her and support her but I always had the doubt that something was not quite right, but the focus on her health was much more important than other things.

Speaking with teachers, I shared with them my doubts and concerns regarding Martina and her academic development, but I always received the same thoughts from her teachers, that Martina will write and read well over time, they also explained Martina is not in the classroom most of the time, she has to go out to receive her therapies, medications and misses out a lot of what they do in the classroom, but despite all that, Martina was fine.

What can you do in that case? I only trusted the school because they are the teachers, the ones who studied for this and I did not want to go against it, I even ignored my opinions as an early childhood educator myself, I always chose to agree with them because I trusted that they had greater and updated knowledge in these situations.

Time went on and Martina continued her classes, everything became even more difficult for her and for me, I was the one who tried to help her at home with her homework. We reached a point where my husband did not want to waste more time and he made the decision of doing evaluations on Martina through a pediatric neuropsychologist, to be sure that there was nothing else going on. I continued to trust the words of teachers and it seemed to me that it was not necessary to do more evaluations and put more stress on my daughter, who was already going through a lot, but I agreed to have these evaluations done.

Well, the diagnosis we received was surprising, “Dyslexia” a 11-years old girl with a 2nd grade level in reading and writing, after hearing this all the pieces in my mind were in their place, I finally understood why it was so difficult for Martina to read, write and solve mathematical problems, since in my years of study to be an early childhood teacher I studied how to
recognize flags of learning disorders, which was exactly what I thought, and at that time, I decided to keep quiet instead of clarifying this in front of the teachers and asking for the corresponding evaluations to find out this type of disorder. I felt it was my fault for not speaking on time and trusting so much in those who were in charge of my daughter’s education, and thanks to my husband because he did not give up until he found the correct professional to do these tests on Martina. After all this, we presented the diagnosis to the school, but we did not receive positive responses from the teachers or the school, they did not believe or did not want to believe Martina had Dyslexia. Therefore, refusing to change methodologies and help her to emerge academically in a different way than the methods they were using, for them, all that was offered, was the necessary, and there were no other resources left to use.

We had no other option to start an appeal process through the NYC Department of Education for the correct services that our daughter needed and the remediation help for the lost years without realizing that Martina needed another form of education to be able to move forward in her academics. We received legal guidance to navigate NYC’s DOE appealing process. The appealing means bring our case to an impartial hearing. In there, an officer will hear both parties in disagreement and give a solution.

In our case, we presented the evaluations and diagnosis from the neuropsychologist about dyslexia as proof of her struggle in school. Also we showed she was left academically behind besides she was in the grade that corresponded to her age, plus all the evidence we had supporting why a teacher that is trained and have the appropriated settings to implement Orton-Gillingham methodology makes a difference in a child with dyslexia, after a long and tedious process everything worked in our favor, Martina now enjoys school because she has what she needs, a better education and all her services, she is being much more independent when doing schoolwork and in general. The approach to help children with dyslexia was developed, compiled, and published in early 1930s.

Orton Gillingham is the name of the methodology, as parents, is hard to understand that not every teacher is trained to detect and distinguish these flags and refer these cases for more evaluations and hopefully the school district can provide support with the correct training to teachers for these circumstances.

It is so comforting to see your daughter can get ahead with the resources she really needs despite the fact the disease already puts so many limitations on her, this left me a very important lesson and I share with other parents who might be seeing their children are not progressing at school as they supposed to, perhaps they are not really receiving the help they need, we must ask for the necessary conditions so that our children can move forward.

And yes, anything is possible, nothing is impossible, there are ways to support them even more than we do as parents, so they can cope with a very difficult disease that affect so many levels.

The message, speak up for your kids!

CRN and eBay for 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.

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.
As VP of Development one of my duties is to establish, nurture, and grow our relationships with the many and varied industry partners that CRN works with. I am ever-humbled by the generosity of our partners, but especially to Horizon Therapeutics, Leadiant Bioscience, Recordati Rare Disease, and AVROBIO who all share the same conviction that every person with a rare disease has the right to the best possible treatment. Through their combined efforts, we have secured over $280,000 in funding for the 2021 fiscal year! We collectively say a great big “THANK YOU” to our friends working as tirelessly as we do!

While we are sad at the realization we will not be able to meet in person this year, there has been no slowing down the incredible industry partnerships and associated projects we are currently cultivating. Projects such as PCs for People (which has to-date supplied 38 families/patients in need; high-quality refurbished desktops, laptops, internet and accessories at absolutely no cost!) Being lead on this project, I personally oversee all processes from the inclusion questionnaire to final asset ordering and subsequent shipment direct to your door. The process we have developed is very turnkey and simple, all you need to do is reach out to me directly at jdicks@cystinosis.org and I’ll get working for you!

We are also very proud to announce the brand new addition of interpreting services for our cystinosis community. Language should never be a barrier to seeking healthcare, education or important daily services. In an effort to eliminate these language barriers, Bromberg & Associates has created a no-cost program, sponsored by Horizon Therapeutics, to offer translation and telephonic interpretation for Spanish-speaking individuals and families impacted. Please see program details in English and Spanish on pages 26-27.

Other pipeline projects taking shape include a digital factsheet developed especially for the CRN by the American Kidney Fund that gives best practices and tips for successfully navigating what could be new territory for some in the world of telehealth physicians visits. Be on the look-out
This past March has seen the CRN partner with the Center for Chronic Illness, which offers free, professionally-facilitated support groups and health education programs for those impacted by all types of chronic illness, including cystinosis. We heard what the community was asking for and have developed a totally separate monthly group specifically for adults who are parents of children affected by cystinosis. We envision an open forum where parents can attend to share, learn, and connect all while being led by a professional social worker.

In recent fundraising, we have seen Andrea Carr and crew working incredibly hard the entire month of May (and many months of preparation beforehand as well) for the first annual ‘Miles for Moose 5.7 Mile Virtual Walk/Run’. The event launched May 1st, 2021 to acknowledge Cystinosis Awareness Day and the daily challenges little Benson (as known as Moose) bravely faces each day. Thank you to our Miles for Moose founders and supporters on a successful first event!

Snapshots from the Miles for Moose 5.7 mile event. Moose, also known as Benson, is featured in the upper left photo.
Bromberg’s Translation and Telephonic Interpreting Services Program, sponsored by Horizon Therapeutics

Language should never be a barrier to seeking healthcare, education or important daily services. In an effort to eliminate language barriers, Bromberg & Associates has created a no-cost program, sponsored by Horizon Therapeutics, to offer translation and telephonic interpretation for Spanish-speaking individuals and families impacted by specific health conditions. If you or your family member has been diagnosed with any of the following conditions, you qualify for document translation and interpreting services by telephone by the Bromberg & Associates team at no-cost:

- Rare metabolic conditions (Urea Cycle Disorder, PKU or other)
- Rare kidney disease (Cystinosis, Fabry or other)
- Uncontrolled Gout
- Primary Immune Deficiencies (Chronic Granulomatous Disease, Hyper IgM Syndromes or other)
- Graves and/or Thyroid Eye Disease

To enroll in the program, please complete the HIPAA form (available here) or email Translator@BrombergTranslations.com to have the form emailed to you. That will allow Bromberg to provide services to you. Remember the services are available to you at no-cost.

If you have questions, please call (844) 405-1866 and enter the PIN# 200 or email Translator@brombergtranslations.com. Once you fill out and sign the form, please email it to Translator@brombergtranslations.com.

To obtain translation and telephonic interpreting services, please read the following options:

**OPTION 1: Connect with an interpreter by telephone:**

Once you submit the HIPAA form, you will receive the instructions to connect to a toll-free phone line which will allow on-demand access to a professional interpreter that can assist you with healthcare encounters, emergency calls, educational meetings, depositions and court hearings, and calls to government agencies.

**OPTION 2: To obtain a document translation:**

Once you submit the HIPAA form, scan and email your document to Translator@BrombergTranslations.com. Bromberg & Associates will review and respond with our timeframe to complete your request. Examples of documents you can send for translation are personal documents (birth and death certificates, marriage licenses, academic records, passports and driver's licenses), medical records, forms and applications to government agencies and insurance companies.

Please note that interpreters and translators cannot provide any legal or medical advice or opinion. Interpreters and translators' roles are limited to facilitating communication between English and Spanish and offering cultural clarification.

This program is run and managed by the Bromberg & Associates team and made possible thanks to the generous support of Horizon Therapeutics. All personal information is securely and confidentially stored by Bromberg & Associates. Horizon Therapeutics receives no personal information.

C-HZN-00329
Programa de servicios de interpretación telefónica y de traducción de Bromberg patrocinado por Horizon Therapeutics

Los idiomas no deberían ser barreras para buscar atención médica, formación o servicios cotidianos importantes. Con el ánimo de eliminar las barreras lingüísticas, Bromberg & Associates ha creado un programa gratuito, patrocinado por Horizon Therapeutics, para ofrecer traducción e interpretación telefónica a personas y familias de habla hispana afectadas por determinadas enfermedades. Si usted o un miembro de su familia ha sido diagnosticado con alguna de las siguientes enfermedades, usted puede recibir los servicios de traducción y de interpretación telefónica del equipo de Bromberg & Associates de forma gratuita:

- Enfermedades metabólicas poco comunes (trastorno del ciclo de la urea, PKU u otros)
- Enfermedades de los riñones poco comunes (Cistinosis, Fabry u otras)
- Gota no tratada
- Inmunodeficiencias primarias (enfermedad granulomatosa crónica, Síndrome de Hiper-IgM u otros)
- Enfermedad ocular de Graves y/o tiroides

Para inscribirse en el programa, complete el formulario de HIPAA (disponible aquí) o envíe un correo electrónico a Translator@Brombergtranslations.com para que podamos enviarle el formulario por correo electrónico. De esa forma, Bromberg podrá brindarle servicios. Recuerde que los servicios disponibles son gratuitos.

Si tiene preguntas, llame al (844) 405-1866 y marque el PIN #200 o envíe un correo electrónico a Translator@Brombergtranslations.com. Una vez que complete y firme el formulario, envíelo a Translator@Brombergtranslations.com.

Para obtener servicios de interpretación telefónica y de traducción, por favor lea las siguientes opciones:

**OPCIÓN 1: Contactarse con un intérprete por teléfono:**

Una vez que envíe el formulario de HIPAA, recibirá las instrucciones para conectarse a una línea telefónica gratuita que le permitirá acceder a un intérprete profesional que puede asistirle en encuentros sanitarios, llamadas de emergencia, reuniones educativas, declaraciones y audiencias judiciales, y llamadas a organismos gubernamentales.

**OPCIÓN 2: Para obtener una traducción de un documento:**

Una vez que envíe el formulario de HIPAA, escanee y envíe su documento por correo electrónico a Translator@BrombergTranslations.com. Bromberg & Associates lo revisará y responderá con nuestro calendario para completar su solicitud. Algunos ejemplos de documentos que puede enviar para su traducción son documentos personales (certificados de nacimiento y defunción, licencias de matrimonio, expedientes académicos, pasaportes y licencias de conducir), historias clínicas, formularios y solicitudes a organismos gubernamentales y compañías de seguros.

Por favor, tenga en cuenta que los intérpretes y los traductores no pueden proporcionar opinión o asesoramiento jurídico o médico. Las funciones de los intérpretes y traductores se limitan a facilitar la comunicación entre el inglés y el español y ofrecer información cultural.

El equipo de Bromberg & Associates dirige y gestiona este programa, que es posible gracias al generoso apoyo de Horizon Therapeutics. Toda la información personal es almacenada de forma segura y confidencial por Bromberg & Associates. Horizon Therapeutics no recibe información personal.
January 14, 2021

Dear CRN,

Please accept this donation on behalf of our family and in memory of our daughter, Livia. Our organization, Livgracefully was founded in memory of Mason’s sister and our hope is to help raise awareness for Cystinosis and give back to the community she so loved.

Livgracefully recently held an apparel fundraiser, where our goal was to raise enough funds so we could increase the amount of scholarships we give in our local community to high school seniors who have faced a health hardship or diagnosis. With this fundraiser, we were also able to raise funds to contribute to CRN and the other organizations we have chosen to help.

Thank you for all the work our beloved Cystinosis community continues to do for our loved ones.

Blessings to you all,
Livgracefully’s Board of Directors

and

The Stilke Family (Dave, Kirsten and Mason)
Parenting Cystinosis Support Group

A web-based peer support group for parents of children living with the rare disease, cystinosis facilitated by Kerry Heckman, MSW, LICSW

3rd Tuesday of every other month
(March 16th, May 18th, July 20th, Sept 21st, Nov 16th 2021)
2:30-3:30pm (PST)

To sign up, visit www.supportgroupscentral.com/CCI

Contact us at info @thecenterforchronicillness.org
or (425) 296-2705 with questions
www.thecenterforchronicillness.org
This program is free of cost.
Myrtle Beach Golf Tournament Fundraiser

By Gail Potts

Fundraising is a vital part of the Cystinosis Research Network to meet our mission of supporting researchers, families and educating public and professional communities. Rare diseases lack government funding thus the need for parent organizations to help in this struggle to find better treatment and a cure for cystinosis.

My daughter, Deanna Lynn, was diagnosed in 1973 at 10 months of age with cystinosis. She was one of the pioneers in the development of oral cysteamine treatment and eye drops at the NIH. She lived a life of challenges but remained active in dancing, school activities, and scouts. She underwent dialysis and two kidney transplants. She was a very caring and generous person. At the age of 27 she passed away. Her wish was to help others with cystinosis further their education, knowing the stumbling blocks that are encountered along the way. Thus, a scholarship was established in her memory. Applications are available on at www.cystinosis.org.

Her passing put me on a “sabbatical” from the cystinosis community for a number of years until I was approached and asked to serve on the CRN board. As a member of the Board, I have committed to helping to fund raise. This has led me to step out of my comfort zone and plan a maiden voyage on this new adventure.

I will be co-sponsoring a Golf Tournament fund raiser to support CRN with MM Golf and Travel on August 14, 2021 in Myrtle Beach, South Carolina. It will be a Captain’s Choice format, with a tee off at 8:00 AM. There will be prizes drawings and raffle drawings. Lunch will be provided. There are many ways to support this fundraiser. You can register as a player, be a Hole Sponsor, donate items for the raffle, or become a volunteer. To be a player or Hole Sponsor go to www.MMgolfandtravel.com to register. If you would like to contact me, please email gpotts47@yahoo.com.

Cystinosis Memorial Fund

By Megan Morrill

The Cystinosis Memorial Fund (CMF) has been busy awarding scholarships to teens and adults living with cystinosis to help cover technology, career, and education expenses. So far we have awarded four individuals with $1,000 USD scholarships! The awardees are Emily Patterson, Cheryl Simoens, Niall Barron, and Melina Castro. Congratulations!

The Cystinosis Memorial Fund has more scholarships to award, so keep submitting those applications!

Apply today at https://cystinosis.org/cmf.

Ways to Stay In Touch

CRN Facebook public page @CystinosisResearch or facebook.com/CystinosisResearch
CRN Facebook closed group at facebook.com/groups/6382741905

Twitter @CystinosisCRN

Instagram @cystinosisresearchnetwork

Instagram closed group for teens @CystinosisTEENS

TikTok @cystinosis57
PROCYSBI is available in tear-open packets in addition to capsules

The medicine inside PROCYSBI packets is the same exact medicine inside PROCYSBI capsules.

Available capsule strengths: 25 mg and 75 mg

Available packet strengths: 75 mg and 300 mg

For more information, visit PROCYSBI.com.

To learn about the support services available, visit PROCYSBI.com/Cost-Savings-and-Support.
Now Available - Book Comprised of Cystinosis Stories

A few years ago, Amanda Buck (cystinosis caregiver) and Amanda Leigh (adult living with cystinosis) recruited people impacted by our rare disease to share their stories. The result is a beautifully orchestrated compilation of their perspectives in a book titled, “Strength: Lives Touched By Cystinosis.” It is currently available through amazon.com with proceeds benefitting the Cystinosis Research Network. Thank you to the Editors and each contributor for being a part of this project and sharing your lives with the world.

Hosting a Facebook Fundraiser

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 2.85 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. These often small acts of kindness add up, creating funding for research and support programs. Did we mention, Facebook does not charge a processing fee. This means 100% of your contributions go directly to the Cystinosis Research Network.

How to Get Started:

Here’s how to get started:
Visit facebook.com/fund/CystinosisResearch and follow the prompts. Another option is to follow these instructions:
1. Click 📚 Fundraisers in the left menu of your News Feed. You may need to click See More.
2. Click + Raise Money.
3. Under Nonprofit select Cystinosis Research Network
4. Choose a cover photo and select the fundraiser details
5. Click Create.

Thank you for your continued support!
We are happy to introduce our newest members of ALAB (Adult Leadership Advisory Board) Briana Dundon, Christina Morris, Hannah Creel, and Gracie Smith. They are a welcome addition to our group.

Our podcast series, "Cystinosis Rare 'A Journey into the Unknown," recently launched our 6th episode: "Chronic Illness, Stress and Anxieties During the Pandemic". The episode includes a conversation with Kerry Heckman MSW, LICSW about dealing with mental and emotional issues as related to living with a chronic illness during the pandemic. We invited Kerry Heckman to be our guest because she is a part of the Center for Chronic Illness and has a web-based peer support group for those with cystinosis.

All ALAB podcasts are available on the Anchor app and the CRN YouTube channel.

CystinosisTEENS Instagram

By Megan Morrill

The CystinosisTEENS Instagram was created just for teens and young adults with cystinosis. The Instagram account is doing well with ongoing spotlight posts highlighting teens and young adults’ successes and interests, allowing followers to connect and be reminded that they are not alone with their illness. The account also provides promotional posts focusing on ALAB’s project updates and CRN opportunities for the entire cystinosis community.
Cystinosis Sessions

By Brian Ensor

Cystinosis Sessions is a monthly face-to-face discussion with adults with cystinosis on Zoom. We learn by sharing stories. We do try and stay current with cystinosis and world news, such as COVID-19 and the eye drop situation. It varies regionally though, many times we learn news during the Session. Visit the ALAB Facebook page or cystinosis.org/ALAB to learn more.

Farewell Letter from Outgoing ALAB Chairperson, Cheryl Simoens

It is with both a happy and heavy heart that I write this farewell as I am stepping down from my various roles with the Cystinosis Research Network (CRN) and Adult Leadership Advisory Board (ALAB) to pursue my educational goals. Over the past 2 years, I have developed and honed many skills including creativity, leadership, and communication from my roles as Chairperson and Adult Patient Representative. I facilitated meetings, strategized over various marketing campaigns, participated as a Keynote speaker during the Cystinosis Ireland Conference, and generally put my efforts into providing awareness for cystinosis and educational content for the entire community.

One of the projects that I committed my time and efforts to and am most proud of is the podcast, Cystinosis Rare: A Journey into the Unknown, founded by a lovely team consisting of a few other ALAB members. We worked extremely hard over the years to engage community members by providing a platform for their participation during episodes, ensuring their voices were heard. We also engaged medical professionals and experts in many areas to bring relevant and real content to our episodes for those living with cystinosis. I am proud of the success of this podcast and that it will continue for others to enjoy.

However, as meaningful as everything I mentioned and more has been, I will most deeply cherish the people I have met and friendships I have made. There are such amazing individuals living with cystinosis who have shared experiences and are truly inspirational, warm, and considerate. I felt as though I walked into a community that made me feel so welcome with their words and smiles and created immediate bonds. This I will never forget. We are a very small community, kindness and understanding are vital to the integrity of those inside of it.

I am embarking on a new journey in the fall to begin a master’s program in counseling psychology with intentions of remaining a volunteer and resource for both the CRN and ALAB. I also plan to provide my support and services to CRN upon graduation in a volunteer professional capacity and continue my presence within the community. Please remember to be kind to each other and listen first. You never know who you can help by simply reaching out and talking until you just do it.

Cheers,
Cheryl Simoens
In Memoriam - Stephen G. Webster
September 1, 1974 – September 6, 2020

By Bruce Webster

Stephen was born in Huron, South Dakota. Huron is a long way from any medical expertise. However, we got lucky. Stephen was diagnosed with cystinosis at 15 months. His story in those early years was the typical struggle of a cystinotic and his parents. He was enrolled in the Vitamin C study for a year or so. When that study was discontinued, Steve was enrolled as one of the Pioneer 12 at National Institutes of Health in their cysteamine study. Despite three and a half years of cysteamine, his kidneys started to fail. On March 23, 1984, at age nine, he received a kidney. Receiving a kidney was one of his miracles. Unfortunately, Steve got sick, suffered rejection, and University of Minnesota was one day from taking it out. Just. One. Day. Fortunately we got a reprieve as they were able to treat his rejection successfully. After 50 days at the University of Minnesota, we got to go home to Lincoln, NE, where we lived at the time. (He sure did love his Cornhuskers.)

With his new kidney, he graduated elementary school and we moved to Nashville, TN. He graduated junior high, got his driver’s license, and graduated from high school. Eight years and counting on a Just. One. Day. kidney.

Steve was able to get a job at Pizza Hut, went to Community College and graduated, and went to Middle Tennessee State University (MTSU) and graduated with a Mass Communications degree. That’s 16 years and counting on a one more day kidney.

Living in Nashville, Steve fell in love with country music and loved to don his boots and cowboy hat and go dancing. He developed a love of NASCAR auto racing, a true Southern pastime. He went back to MTSU and earned a second bachelor’s degree in Business Administration in 2010. That’s 26 years for a kidney they almost took out.

After 30 years, his kidney failed and he had to go on dialysis. During his final 6 years, he became very strong in faith and spirituality. He had a gift from God to relay messages and to see visions. He said he had talked to his former minister who had died of cancer, and the minister told Steve everything was going to be okay. Another time he told us that he had talked to Myrtle Ruth, a member of our church who had died of Alzheimer’s, and she said her memory was restored, and to tell Bud, her husband, that everything is going to be all right.

Steve almost got a chance at a second kidney when they discovered colon cancer. They successfully removed the cancer, but he was put on hold for a transplant. That was too much to bear, and with other complications from being in the hospital, he lost hope. He had one last vision of Jesus and both sets of grandparents discussing and voting on whether Steve should stay in this life, or come to his heavenly home. The next day, his 46th birthday, he quit treatments, medications, and dialysis, and spent his last days at home.

Steve was born the day before Labor Day, and he died the day before Labor Day. He was never married, and was deeply lonely at times. But he is now at peace; he is now healed; and is now in his heavenly home with his Lord and Savior Jesus Christ. He no longer struggles with medical issues and problems, and no longer has to live under the cloud of Just. One. Day.
In the late 1970’s, a hospitalization led to a cystinosis diagnosis which was uncommon at the time. Karen Gledhill, and ultimately her sister, discovered they had late onset cystinosis in their 20’s. During this year’s Cystinosis Awareness Day efforts, Karen shared an original letter written by her mother, providing a glimpse into their journey.

Dear Friends,

Please bear with me while I try to chronicle the events of the past 64 years. In a way, it is a bit complicated because two of my children are involved. Also, it is not the typical case of cystinosis that you may be familiar with.

Instead of symptoms developing in infancy, early childhood or teen years, my daughters were approximately twenty-five years old when their trouble became noticeable. Dr. Schneider hadn’t encountered any patients in this age group before our family was brought to his attention. He wondered if he should call it "Late-Late Onset".

My husband and I live in Rochester, New York and are the parents of five children; Karen - age 31, Kathleen - age 29, Debra - age 26, Daniel - age 18 and Thomas - age 17. Karen and Deby have Cystinosis. Karen is married and works in an advertising agency. Deby moved to California seven years ago. She is single and works in Beverly Hills.

Karen was hospitalized in the fall of 1977 for test when she was found to have kidney problems - a complete surprise to all of us. Four attempts at a needle biopsy were made and all were unsuccessful. Some tissue was obtained but it was only scar tissue and no conclusion could be reached as to the cause of her diminished kidney function. Therefore, she was treated as a regular kidney-disease patient. By August 1981, it was necessary for her to go on dialysis. Her doctor was associated with a self-care unit and Karen learned to handle the dialysis machine and monitor herself while on it.

When we found out Karen was headed for dialysis, it was decided to test the older members of the family to see if anyone would be compatible enough to donate their kidney to her. To make a long story short, those whose blood type matched (Kathleen and myself) were found to have traces of blood and "casts" in our urine and were eliminated. My sons were then given routine urinalysis tests and the same thing showed up in them. Needless to say, we were very upset because we didn't know if this meant kidney problems for our other children too. Dr. Rufino C. Fabico, Nephrologist at Strong Memorial Hospital became involved with our family at this time. He keeps track of us through routine urinalysis and blood tests twice a year.

Back in Los Angeles, about two months later, Deby wasn’t feeling well and went for a physical. Her urinalysis test showed only 40% kidney function and other irregularities plus her blood pressure was quite high. She was referred to a specialist who had her hospitalized in order to do a kidney biopsy. The needle biopsy was unsuccessful in Deby’s case. However, because of the family’s medical history, her doctor ordered a surgical biopsy. We were fortunate that the pathologist who examined the tissue was alert and found the cystine crystals for they could have been washed away during normal biopsy procedures.

We were then told Deby had "Cystinosis". What in the world was "Cystinosis"? Her kidney doctor didn’t even know for sure and had to get more information on it. Supposedly, only "little kids" get it. Why did it show up in our family and why did it show up when it did? Dr. Fabico was a tremendous help to us at that time. He had written some papers and articles on Cystinosis after treating another family. He explained this disorder and how it affects the body. We still don’t know why it developed so late in our daughters.
CRN has funded over $5 million total in research grants and fellowship, including a cystinosis fellowship at the National Institutes of Health, research and education programs in the United States and many countries around the world including Egypt, Mexico, England, Scotland, Italy, Belgium, France, and much more. CRN has also co-funded research projects with Cystinosis Ireland. CRN research topics have focused on every aspect of cystinosis with the purpose of understanding the disease and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, fertility, improved medications, psychological and much more. CRN’s current grant commitment is over $600,000.

**Current CRN Grant Commitments**

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

**Cognitive Control Systems in Cystinosis**

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

Grant Amount: $315,193, two year study

Seminal behavioral studies by Trauner and others in human patients have suggested the presence of cognitive dysfunction in cystinosis. Our own work suggests some behavioral and neural differences and difficulties with sensory memory in this population. However, the neurocognitive phenotype associated with CTNS mutations and its developmental path are still poorly understood, and the prime areas of neurocognitive vulnerability in this population are in need of much more thorough characterization. This is critical to developing effective therapies to compensate for or improve on areas of cognitive vulnerability. To this end, we propose to characterize different components of executive functioning (memory updating, set shifting, conflict monitoring, and inhibition) in cystinosis. Focus on this area is motivated by our previous work, the literature on cognitive weaknesses in cystinosis thus far, and by first person reports collected during interactions with patients and families. We will use high-density electrophysiology (EEG)—a non-invasive method that allows one to directly measure functional brain activity at the millisecond scale and thus reliably assess the integrity of information processing at the neural level—and standardized cognitive functional assessments to test 15 children, 15 adolescents, and 15 adults with cystinosis and the same number of age-matched unaffected healthy controls. Twenty heterozygotes and age matched controls will also be tested to examine the role that mutation versus disease plays in the cognitive phenotype of cystinosis.

Significance: Greater knowledge of the neurocognitive dynamics of executive function in cystinosis has the potential to critically advance the development of interventions for these individuals. In addition, identification of neural markers will provide objective assays of treatment efficacy on brain function.

**Chitotriosidase as a Therapeutic Monitor for Cysteamine Therapy**
in Cystinosis: a Retrospective Validation Study

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

Grant amount: $44,000, one year study

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

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis

Katharina Hohenfellner, MD

Grant Amount: $77,000, one year study

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

Potential Impact for patients with Cystinosis:


Grant Awarded February 2021 by the Cystinosis Research Network and Cystinosis Ireland

Perturbations in the V-ATPase Pathway Drive Pathology in the Male Reproductive System in Cystinosis

Principal Investigator Professor Minnie Sarwal, Professor of Surgery, Division of Multi Organ Transplantation, University of California San Francisco (UCSF), USA and co-applicants, Dr James F. Smith, Associate Professor and Director Male Reproductive Health, Department of Urology, University of California, San Francisco and Dr Polina V Lishko, Associate Professor, Department of Molecular and Cell Biology, University of California Berkeley, USA

The research project is a total investment of €300,000 from Cystinosis Ireland and CRN (€150,000 each) over the next three years.
Cystinosis is a very rare inherited genetic disease that causes the build-up of cystine, an amino acid normally present in very small amounts in every single cell of a healthy person. The excess cystine forms sharp crystals that damage the body’s cells. Many of the body’s organs are affected by cystinosis including the kidneys and the eyes in particular. However in men, there can be an impact on fertility and the ability to produce sperm (azoospermia). Whereas in the past, the life expectancy of men living with cystinosis was short and their physical wellbeing relatively poor, today there are an increasing proportion of men living with cystinosis who are well and who want to consider parenthood. This research project aims to study the molecular and cellular changes that can cause azoospermia in men with cystinosis. The research will be a first step towards developing an effective treatment that will give men living with cystinosis the opportunity to become fathers. The knowledge generated from this research will also improve our overall understanding of the disease and in particular of certain poorly understood cystinosis symptoms that appear to be caused by malfunctions other than the accumulation of cystine. In selecting this proposal for co-funding, the Boards of Cystinosis Ireland and CRN agreed that this is a scientifically significant proposal focused on a very important and strategic research topic for cystinosis patients.

This project builds upon research and results generated from two previous projects co-funded by Cystinosis Ireland and the Irish Government’s health research funding agency (the HRB) – a project led by Professor Minnie Sarwal in UCSF, USA entitled “Targeting Autophagy in Nephropathic Cystinosis” and a project led by Professor Elena Levchenko in UZ Leuven, Belgium entitled “Unravelling the mechanisms of azoospermia and potential future treatments in male cystinosis patients”.

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

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

A Cellular Resource for Studying Male Infertility in Cystinosis, Minnie Sarwal, MD, PhD, Professor of Surgery, (Director, Precision Transplant Medicine) University of California San Francisco (UCSF), James Smith, MD, MS (Director, Male Reproductive Health Center Urologist), University of California San Francisco (UCSF), Ann Harris, Professor, Department of Genetics and Genome Sciences, School of Medicine, Case Western Reserve University, Cleveland, Ohio, Elena Levchenko, Professor, Department of Pediatric Nephrology, Leuven, EU, and Swastika Sur, MSc., PhD Postdoctoral Scholar, Sarwal Lab, University of California San Francisco, Department of Surgery.

Total Grant: €10,000

Principal Investigator, Swastika Sur, a Postdoctoral Fellow at the University of California San Francisco, has outlined a research proposal focused on infertility in men with cystinosis. In addition to various endocrine organs that are affected in cystinosis, hypergonadotropic hypogonadism has been reported as a frequent finding in male cystinosis patients. Although spermatogenesis has shown to be intact at the testicular level in some patients, no male cystinosis patient is known to have naturally fathered a child. The sole treatment for cystinosis is the aminothiol cysteamine, which is highly effective in reducing the intracellular levels of cystine. However, this drug is ineffective in the treatment of male infertility in these patients.

A previous study of the pathophysiology of cystinosis-mediated infertility used a CTNS-/- mouse model. However, the CTNS-/- mouse model generated on C57BL/6 background was found to not be suitable for clarifying the pathogenesis of male infertility in cystinosis. This mouse model partially mimics the renal phenotype of the human disease but was found to have normal testicular morphology and function. Therefore to this date, the exact pathophysiology of male infertility observed in patients with cystinosis is not yet fully understood, which is a critical unmet need due to the growing population of cystinosis patients, treated with cysteamine reaching young adulthood.

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

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

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

The Specific Aims of this project are the following:

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

· Aim 2: Map the molecular perturbations in both cell lines with deletion of CTNS and in tissue samples from male cystinotic patients, by using state of the art genomics that the Sarwal Lab has legacy expertise-in. This will define the clinical utility of the resource generated in Aim 1.

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

**Cystinosis Community Advisory Board**

I continue to participate as the U.S. representative in the Cystinosis EuroCAB programme, a project of EURORDIS, the European Rare Disorder Organization. The Community Advisory Board’s (CAB’s) objective is to improve patient access to novel therapies and treatments. This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research. The CAB has been quite active in 2021, meeting with two industry sponsors in April and May. We look forward to continued partnership with researchers and industry worldwide to improve the quality and speed with which
cystinosis treatments are developed with the patient’s voice in mind.

**National Institutes of Health**

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

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

**Educational Resources**

All of CRN’s educational materials including brochures, guides and other publications have been updated and are available on the CRN Website. An expanded Dialysis and Transplant section has been created to include a broad range of information and resources for those facing these challenges at [https://bit.ly/3mFzzy7](https://bit.ly/3mFzzy7).

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://bit.ly/35nIYnN](https://bit.ly/35nIYnN).

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

*By Jenni Sexstone, Treasurer*

For the 3 months ended March 31, 2021

**Revenues**

Total income for the three months ending March 31, 2021, was $149,000 compared to $294,000 in 2020 due to timing of grant receipts.

**Expenses**

Total expenses of $159,000 were higher than expenses for the same period during 2020 of $32,000. In the current fiscal year we have funded $138,000 of grant payments for research. All other expenses were $11,000 lower than the same reporting period in 2020 due to timing of newsletter expenses.

CRN had net operating loss of $10,000 for the three months ending March 31, 2021. Continuous direct public donations, fundraising activities and generous corporate support continues to provide cash resources to increase patient advocacy and family support activities in 2021 and beyond to support the cystinosis community. Cash on hand at March 31, 2020 was $511,000. Net change in cash for the first quarter 2021 was a decrease of $7,000.

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**Scholarships Available for Collegiate or Vocational Program Students**

We are pleased to be able to continue our commitment to our community by providing the following scholarships:

- Individual Living with Cystinosis Scholarship
- Sierra Woodward Sibling Scholarship
- Deanna Lynn Potts Scholarship

Each of these scholarships offer an award of $1,000. Applications and information for applying are available on our website at [cystinosis.org/scholarships](http://cystinosis.org/scholarships). We are now accepting applications. The deadline is August 1, 2021. If you have questions you can contact Gail Potts at gpotts47@yahoo.com.
At Leadiant, we understand that those living with cystinosis have needs that extend far beyond medicine. We are committed to providing educational resources to support the management of eye health throughout your lives.

Our Rare Dedication to the cystinosis community is stronger than ever. We have been with you for over 20 years – through great progress and challenges – and we will remain with you.
TRAUMA-LESS NEEDLE POKE WELLNESS KITS

ANGEL AID CARES IN PARTNERSHIP WITH MIGHTY KIDS CAN ARE OFFERING FREE WELLNESS KITS FOR RARE DISEASE PATIENTS.

Sponsored by HORIZON

Needle-poke procedures can be stressful and sometimes scary for patients. To help these patients and caregivers we’ve created the Trauma-Less Needle Pokes Wellness Kit. Each kit contains pain management TOOLS, DISTRACTION CARDS, and SELF-CARE AIDS to help patients get through their needle pokes with less pain and anxiety.

Your FREE Wellness Kit will include:

- Trauma-less Needle Pokes 7 Steps Checklist
- Buzzy® XL Bumble Bee XL pain-blocker device pack, including: 2 ice wings, 1 comfort strap, batteries
- Buzzy DistrACTION® cards
- Bravery Stickers
- Stainless steel water bottle + Nuun hydration supplements
- Tactile soothing object
- Free access to meditation app

*Items subject to change based on availability. Kits are reserved for patients in the CGD, UCD, and Cystinosis communities. Others will be put on a waitlist. First come, first serve. While supplies last. Kits will be shipped on a monthly basis starting at the end of February. Questions? Email Programs2@AngelAidCares.org

Request a Kit, It's FREE!
www.angelaidcares.org/mighty-kits

This kit works in conjunction with the on demand webinar, Trauma-Less Needle Pokes: 7 Steps to Easier Injections, Blood Draws, and IVs. Watch the webinar at www.angelaidcares.org/needle-pokes
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.
## 2020 Donor Honor Roll

### $100,000+
- Horizon Pharma USA, Inc.
- Leadiant Biosciences, Inc.

### $50,000-$100,000
- Shared Health Alliance

### $25,000-$50,000
- Sargalski, Camellia Estate

### $15,000-$25,000
- Avrobio

### $2,500-$4,999
- Recordati Rare Diseases Inc.
- Schleuder, Don L.
- Carmichael, Scott and Tia
- Markel Corp. c/o CyberGrants
- Roesler, Pamela and Jeffrey
- Wyman, Jennifer and Tim

### $1,000-$2,500
- Kelly, John F. & Michelle S.
- Bank of America Employee Giving Campaign
- LDC NWTF
- Rollinger, Khristine
- Ryczek, Kristi
- Greeley, Christy & Jack
- American Society of Nephrology
- Exchange Club of Dearborn Foundation Inc.
- Finn, Janice B.
- Geerdink, Edwin & Maria
- Gilberg, Josh & Katy
- Livgracefully, Inc.
- Morander, Jeffrey and Kim

### $250-$499
- Charities Aid Foundation of America
- LeBeau, Brett and Brittney
- Sevel, Randy
- Kaskel, Dr. Rick and Mrs. Phyllis
- Norling, Richard
- Arndt, Richard and Patricia
- Darbee, Elaine & Calvin
- Hammond, Jack and Colleen
- Gilberg, Frederick R. & Beverly A.
- Moore, Clint
- Ballenger, Roger & Mary
- Daley, Gerald and Elaine
- Guda, Swaroopa
- Moeller, Michael F.
- Roberts Family Foundation
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CRN VISION
The Cystinosis Research Network’s vision is the acceleration of the discovery of a cure, development of improved treatments, and enhancement of quality of life for those with cystinosis.

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

Please email any contact corrections to info@cystinosis.org.