

THE CYSTINOSIS Advocate

Inside This Issue

President's Letter	2
Expanded Care Package Program	4
Cystinosis Network Europe	4
Cystinosis Foundation UK	5
Cystinosis Ireland	5
Education & Awareness Committee Update	8
Global Genes Summit	11
Family Support Committee Update	12
Oscar's Story	13
Cystinosis Memorial Fund	17
CRN Board Member Updates	18
Brain Function Research Study	20
Development Committee Update	21
Myrtle Beach Golf Fundraiser	22
Live Like Laura Fun Fund	25
Adult Leadership Advisory Board	26
Rare Disease Family Impact	29
Free Translation Services	37
Research Committee Update	40
Financial Review	44
NORD Assistance Program	46



The 10th biennial CRN conference was held July 16-18, 2021. Providing a fully online format, attendees from around the globe gained access to renowned experts in cystinosis and intimate breakout groups based on personal interests. Some of the more highly attended sessions included an expansive medical panel presentation, CRN welcome/overview/a review of new and old/anticipating kidney transplant track and product and research updates.

The event's success was possible because of our dedicated volunteers and scientific review board/medical review committee, partner companies and participants. We are grateful to PCs for People for providing computers and internet connection to those in need and Bromberg & Associates for breaking down English/Spanish language barriers. (Both programs remain open to the cystinosis community. For details on PCs for People, visit page [44](#). Spanish translation information on page [37](#).)

28
U.S. States Attended



12
Countries Represented



~300
Attendees



2023 CRN Family Conference Host City:
Nashville, TN

Cystinosis Research Network Board of Directors

President

Jonathan Dicks

Executive Director

Christy Greeley

Vice President Development

Jonathan Dicks

Vice President Education and Awareness

Marybeth Krummenacker

Vice President Family Support

Jen Wyman

Treasurer

Tim Wyman

Secretary

Karen Gledhill

Directors

Carol Hughes

Gail Potts

Kristina Sevel

Herberth Siegler

Adult Leadership Advisory Board Representative

Sara Healy

Scientific Review Board Chairperson

William A. Gahl, M.D., Ph.D.

Membership

Frederick Kaskel, M.D., Ph.D.

Craig B Langman, M.D.

Roslyn B. Mannon, M.D.

Jess G. Thoene, M.D.

Medical Advisory Committee

Corinne Antignac, M.D., Ph.D.

Rachel Bishop, M.D.

Donald Cairns, Ph.D.

Ewa Elenberg, M.D., M.Ed.

Patrick Gipson, M.D.

Paul Goodyer, M.D.

Larry Greenbaum, M.D., Ph.D.

Julie R. Ingelfinger, M.D.

Galina Nesterova, M.D., FABMG

Minnie Sarwal, M.D., FRCP, DCH, Ph.D.

Mihir M. Thacker, M.D.

Doris A. Trauner, M.D.

Joshua J Zaritsky, M.D., Ph.D.

Professional Advisory Committee

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



Finn, Jon (center) and Elle celebrating Halloween.

The President's Letter

I sit back and think about where we have been as a family over the last three years and just shake my head and smile. From attending our first family conference, to advocating on Capitol Hill for rare disease week, to VP of Development and now this? Me, writing the CRN President's Letter? What a long, strange journey it has been. Three years ago, we all as a family came to Philadelphia for the 2019 CRN Family Conference as bright-eyed and unprepared as my first shift in the ER as a critical care nurse. I knew there was a job to be done, I just forgot how to work my legs for a moment. I was so overwhelmed and shocked by the pictures I saw, the stories I read, and most notably the people I met. Little did I know how important those three days would be for me and my family, and as a community with such a rich history of fellowship and celebration; this unknowingly would be the last time we would be meeting in person. In the times since, many things have changed, but we as a family have remained true to our mission of taking care of our children with love and grace, laughing more than we cry, and learning to release that which we cannot control.

This year as we celebrated Thanksgiving and with the holiday season in full swing, I am reminded of how much I crave the in-person and face-to-face interactions within our small community. I am reminded how deeply connected we all are, yet how isolated we can make ourselves in times of great stress. As the newly elected President of the Cystinosis Research Network, I thought I would step into this role much as I did three years prior, with a fear of the unknown. Those feelings simply melted away when I realized who this community was and what I could be doing with my own talents to not only offer Elle a path out of her darkest of times, but to be the change that I wanted to see; to be the light in the darkness. We have weathered a stormy year and a half with no guarantees. But as sure as the sun rises every morning, we're going to be there getting meds dialed in, pulling up syringes, blending g-tube smoothies,

putting in drops, giving shots, logging miles as we hit the many specialists appointments and blood draws that ensure my daughter is in the best possible position to thrive. Pandemic or not, cystinosis doesn't quit, so neither will we and neither should you.

This year I am thankful for all of you, the cystinosis community at large. I am thankful for my family, for my children who make my world so rich and interesting, who love me in a singular way that is only reciprocated by a father and mother's undying and eternal love. I am thankful for my incredible staff of volunteer moms, dads, patients, physicians, entrepreneurs, and industry professionals who donate their time and money so that this vision beginning way back in 1996, continues to espouse our dedication to supporting and advocating for research, providing family assistance, and educating the public and medical communities about cystinosis. I am thankful we pulled off a super successful 2021 Virtual Family Conference full of wonderful information with leading edge updates on the continued research progress, and most importantly I got to see all your beautiful smiles.

As we all undoubtedly feel the subtle acceleration of speed, we know the race towards a cure is fast approaching. But we also know an available cure is only realized by playing the "long game" and navigating the ups and downs of waiting, remaining compliant to the regimen. As such we remain committed towards the development of improved treatments and enhancement of quality of life for those with cystinosis, but with a keen eye towards a very bright and hopeful future.

For those newly diagnosed, I know what you're going through and I hear you. I know the feeling of trying to take sips from a fire hose with a straw, fearing you could drown at any moment. For all of you, hear my words, the same words that were repeated to me back in Philly, "...things are going to get better and the impossible days will soon become possible, we are here and ready to catch you." To my veteran warriors out there, my love and honor for you all is indescribable. You and your tenacious parents literally paved the way with grit and determination in times where experimental trials were your only hope. You stepped into the great unknown, and through your sacrifices we are here now.

In remembrance of those beautiful souls who have fought the good fight, who finished their course; your sparkle, your shimmer forever lights our hearts and the hearts of this community. A green ribbon with a twinkling white border... this is who we are. The rare, the determined, the ever hopeful, cystinosis warriors. We here at the CRN are honored to fight with you and for you until cystinosis is a disease of the past.

May this holiday season find you in good spirits and with loved ones. Sending each and every one blessings and love.

Jonathan Dicks

AVROBIO Update

In a quarterly update, on August 5, 2021, AVROBIO announced:

"Data from the patients dosed to date across three indications continue to support our first-in-class, one-time investigational gene therapies as potentially transformative treatments for the more than 50,000 people worldwide living with the life-limiting lysosomal disorders we are researching."

Among the 12-month milestones anticipated for AVR-RD-04 in cystinosis, the company cited:

- FDA granted Fast Track

Designation for AVR-RD-04 and cleared the Investigational New Drug (IND) application for the AVROBIO long-term follow up trial for patients dosed in the investigator-sponsored Phase 1/2 clinical trial¹ of AVR-RD-04 (CTNS-RD-04) led by our collaboration partner at University of California San Diego (UCSD).

- Stephanie Cherqui, Ph.D., principal investigator of the investigator-sponsored Phase 1/2 clinical study, provided an update on the patients dosed to date in the Phase 1/2 trial at the virtual 24th Annual Meeting of the American Society of Gene & Cell



Therapy (ASGCT), May 11-14, 2021.

- AVROBIO expects to provide a clinical and regulatory update in the first quarter of 2022 and is planning to initiate a company-sponsored clinical trial in the second half of 2022, which it believes could potentially serve as a registration trial, subject to regulatory clearance.

For the full article, access the press release here:

<https://bit.ly/AVROnews>.

Now Expanded - Care Package Program

The Care Package Program was created to help individuals and families with cystinosis during trying times. In the past, we've offered free kits for the newly diagnosed, those awaiting/ receiving a kidney transplant and families who've lost a loved one with cystinosis. We are pleased to announce our program has expanded to include a **wellness package**.

Partnering with Emily Mello and Maya Doyle, MSW, PhD, LCSW-R, the wellness kits aim to deliver comfort items and supportive tools. Here's what they had to say about the initiative:

“ The wellness boxes are a good reminder that you are not in this alone. While each person's experiences are unique to them, knowing you are part of a community that shares similar challenges and cares for each other can make such a difference. ”

– MAYA DOYLE

“ We hope the wellness packages spread love and joy to those in our community who need a little extra TLC and cheering up; a reminder to enjoy little things, fight during the tough times and know they are not alone. ”

– EMILY MELLO



Sign up is available today at cystinosis.org/care-package.

Cystinosis Network Europe

By Denise Dunne



Cystinosis Network Europe recently held its first Annual General Meeting where it reported on its activities in 2020 and 2021. You can find the annual reporting on the CNE website – www.cystinosis-europe.eu.

Through the Worldwide Cystinosis Community Advisory Board (CAB) CNE continues to work with a number of pharmaceutical and healthcare companies as well as researchers in the field, to promote the best research and ensure the patient perspective is to the fore of their work. The CAB recently did some in depth work on Patient Reported Outcome Measures (PROM) and Quality of Life (QoL) tools and gave input to the Public Patient Involvement (PPI) element of a funding call Cystinosis Ireland is undertaking.

The Dutch and Flemish Cystinosis Support Group is working hard on the 2022 CNE International Cystinosis Conference which will be held in Leuven, Belgium next July, and will keep the community updated as plans are finalised.

Cystadrops Finally Available for Patients in England

By Will Newman



After over three years of disappointment for the UK Cystinosis community, NHS England have finally agreed to routinely prescribe Cystadrops. Until now, the only treatment available was an unlicensed product that requires constant refrigeration and needs to be administered every waking hour to be effective. Cystinosis Foundation UK and Metabolic Support UK (an

umbrella organisation supporting patients and families affected by inherited metabolic disorders) decided to work in collaboration, pooling and sharing resources to collate patient evidence and present the patient voice. By designing and publishing a tailored patient experience survey, using online platforms, gathering quantitative and qualitative evidence via a combination of face to face

and online engagement, MSUK and CFUK were able to describe the impact of cystinosis on patients and families. Despite this powerful message it has taken three submissions (2019-2021) to NHS England's Clinical Priorities Advisory Group for the medication to finally get the green light. It is hoped that this will improve the chances for Cystadrops to be approved for Scotland, Wales and Northern Ireland.

Further information is available at:

<https://www.cystinosis.org.uk/news/cystadrops-now-available-for-patients-in-england/>

Cystinosis Ireland Update

By Denise Dunne



Cystinosis Ireland and Cystinosis Research UK are proud to announce their first funding collaboration to jointly fund important research into cystinosis to a total value of €250,000. The first research project is entitled

“Analysis and exploration of novel treatment approaches for muscular disease in cystinosis”, which will be led by Principal Investigator Professor Elena Levtchenko. The research project comprises a combined

total investment of €100,000 from Cystinosis Ireland and CFUK (€50,000 each) over the next two years.

The second research project is entitled “Treating ocular cystinosis by contact lenses”, which will be led by Principal Investigator Professor Anuj Chauhan. The research project comprises a combined total investment of €150,000 from Cystinosis Ireland and CFUK (€75,000 each) over the next two years. More details of the projects being funded can be found at www.cystinosis.ie or www.cystinosis.org.uk.



Sharing your journey with **cystinosis** can advance new treatments

Because cystinosis is rare, researchers need more information directly from patients and their families to understand the condition.

Your progression, symptoms and health challenges are all important clues that can help researchers develop new treatments.

How you can help

Join other cystinosis patients and families on AllStripes to contribute to multiple research efforts.

- 1. Go to allstripes.com/cystinosis**
- 2. Create your account**
Sign up, review our research consent and share the names of your health facilities (about 10-15 minutes). No appointments, no uploads.
- 3. Success — you're a hero for cystinosis**



We collect and de-identify your medical records to power research. You'll receive research updates and all your records in a private, secure account, **at no cost**.

What is AllStripes?

As the leading research platform dedicated to rare diseases, AllStripes makes it easy for patients to contribute to new treatment studies from home.

We do the work to collect and analyze your de-identified medical records to help power faster, better drug development for your condition.

To learn more contact us at community@allstripes.com or visit allstripes.com.



MAGISTRO

FAMILY FOUNDATION

February 2021

The Magistro Family Foundation (MFF) is a small, California-incorporated family foundation created in 2002 by Charles M. and Mary Noël Magistro.

The foundation's original Mission was to *“support non-profit activities and organizations in the areas of education, health and human services.”* In 2017 the Foundation's Mission was expanded to include the areas of Arts and Culture and special projects of the Board of Directors. This last category contemplates one-time contributions to worthy 501(c)(3) organizations.

Over the past 19 years, the MFF has distributed over \$7.5 million to 41 different organizations. A large portion of the total given has been to the Foundation for Physical Therapy Research, an entity that our founder, Charles M. Magistro, helped create and nurture.

In 2021 the Magistro Family Foundation will donate funds in the areas of healthcare, education, arts and culture and special projects of the Board of Directors. The Foundation does not accept unsolicited proposals.

Preferences are given to:

- Projects whose goals are measurable, clear, and achievable
- Organizations that work to attract funding from a variety of sources
- Requests which allow organizations or programs to become more efficient and effective

Please note:

- **Organizations must be classified by the Internal Revenue Service as public charities and tax-exempt under section 501(c)(3) of the Internal Revenue Code of 1986.**
- **Proposals are considered annually.**
- **In 2021, the deadline to apply is April 30, 2021. Incomplete proposals will not be considered.**

Feel free to contact us with questions.

Sincerely,

Paulette Simpson, President
Magistro Family Foundation
magistrofoundation@gmail.com

**The CRN is the proud recipient of a \$20,000 grant through this invitation only initiative.
We are grateful to the Magistro Family Foundation for their generosity.**

Education and Awareness Committee Update

By Marybeth Kruppenacker, Vice President of Education and Awareness



It has been and continues to be an extraordinary time we are in. We at CRN continue to move forward with so many opportunities for our cystinosis community and always with an eye on our goal of advocating for new and improved treatments and an eventual cure!

This year has been like no other for all of us and the ongoing COVID pandemic has taken us all in so many different directions. Everyone in the rare disease community has had to adjust to a very different way of delivering messages and doing outreach to their communities and constantly “adjusting our sails” to where we are going. The Cystinosis Research Network is no different. I think the one thing we can say in living with a rare disease is that our lives can change in an instant and I think that is why we as an advocacy organization have had to learn to do the same... change is not a four letter word!

We at CRN continue to provide opportunities to so many and I think we are committed to offering so many opportunities. We have awarded over \$40,000 in academic scholarships over many years and I am always impressed with our applicants and their goals and what the future holds for all of them. This year was no different with our selections: Kacy Wyman, Mason Stilke and Heidi Hughes (pictured here). Congratulations to our 2021 winners and we wish them all success in their future endeavors. We are so fortunate to continue to have these opportunities available to our community and to be able to offer financial assistance. We have heard from some of our previous awardees about what it meant to receive these different scholarships and it is always humbling to me to read that we have made a difference. (Please see their comments at the end of this article)

I think one of the proudest moments for me this year was the first ever CRN Virtual Family Conference. While we can all agree there is nothing like meeting in person with all of our families and nothing will ever replace that but we were able to utilize the skill and talent of so many within our community and outside our community to work together to bring many of our world’s leading researchers to our virtual meeting. It was extraordinary to be a part of a virtual family conference and to hear from and listen to our speakers. The CRN Board worked hard and spent many hours in fine tuning this first time opportunity to give our families “time together” and to hear and ask the questions of our researchers and



to listen to progress being made for our patients and family members. We look forward to once again meeting in person in 2023!

We continue to represent at some of our nationally known organizations and again, while not in person, the virtual format does offer us the opportunity to still be a part of these meetings as an exhibitor. We do look forward to 2022 where the live

meetings will once again be a reality; PAS scheduled right now for Denver and ASN in Orlando. We look forward to maintaining a presence at these professional meetings. In addition, we are continuing our membership with the National Health Council, NORD, EveryLife Foundation Alliance and other groups who help us stay in touch with the rare disease community.

Finally, I am so very proud of the work of our ALAB group (Adult Leadership Advisory Board). They are a grassroots group within our CRN community and have stepped up with their own voices and taken on the role of advocates for themselves. I can't think of better representatives for the cystinosis community! They are always looking for volunteers and have added some new members this year.



“ My name is Kole Binger and I was honored to be a recipient of the CRN Cystinosis Sierra Woodward Sibling Scholarship in fall of 2020 for Medical School and also in the Fall of 2013 while I was attending the University of Wisconsin-Madison. Growing up, I did not anticipate that my family's health situation would have such a profound impact on my professional choices in life. But it has. My brother, Kellen, was diagnosed with cystinosis at the age of 1 and received a kidney transplant 13 years ago from my father. As a sibling, seeing my brother grow up with cystinosis sparked my interest in medicine and motivated me to pursue a career as a physician. It was incredibly humbling to be supported through CRN from the Sierra Woodward Sibling Scholarship as I began my medical training journey. This award helped support my adaptation to attend school online and it also was incredibly humbling to know that I had the support of the CRN community as I began this journey. I am extremely grateful for the Woodward family and all those who work and volunteer at CRN for making this award possible. ”

– KOLE BINGER

“ The scholarship I received from CRN has helped me immensely. The financial assistance was a big stress reliever, but just as important, was knowing CRN was supporting me and had my back in my post secondary endeavours. I will always appreciate it. Thank you so much CRN! ”

– KATHLEEN ROBERTS

SAVE THE DATE!
5th Annual Cystinosis Awareness Day
May 7th, 2022

Join the effort this
Cystinosis Awareness Day.
Email info@cystinosis.org
to get started!



“ Hi CRN community! As a scholarship recipient from CRN as a high school graduate in 2014, the award enabled me to pursue a degree in Global Supply Chain and Operations Management at the University of South Carolina’s Darla Moore School of Business over the next four years. Graduating with Magna Cum Laude honors in 2018, I was offered a position as an Industrial Engineer with the Commercial Airplanes division of the Boeing Company. I have recently surpassed my three-year anniversary with Boeing, where I am lucky to use the degree I received to solve complex and chronic problems across the production system for the Boeing 787 airplane in North Charleston, SC. Thank you again to the CRN community for all you have done, especially for my sisters, Sarah and Kennedy!

– CAROLINE LARIMORE

”

“ I received the Cystinosis Research Network and the Deanna Lynn Potts scholarships in 2011. I used the CRN and Deanna Lynn Potts scholarships to attend Parkland Community College in Champaign, IL. I received an Associate in Applied Science specializing in Veterinary Technology. After graduating Parkland College in May 2013, I took the national exam to become a Certified Veterinary Technician (CVT). I received my first job as a CVT in October 2013 and have been in the profession for the last 8 years. Although my profession as a CVT has had its ups and downs, I still enjoy going to work every day to help educate owners on how to care for their pets. I am very grateful to the CRN and Deanna Lynn Potts Scholarship for offering these scholarships because without it I would not have been able to afford the tuition for Parkland College or been able to pursue the profession I love.

– MIKAELA GARD

”

“ Almost 10 years ago, I was the recipient of a CRN scholarship. The \$1,000 meant the whole world to me and kept my spirit alive believing there is light at the end of the tunnel. CRN and its board members went beyond my expectation. Board members exercised their discretion in awarding me this scholarship to go back to school – I was 30 years old, my only child just diagnosed with cystinosis, no job, home was over 4,000 miles away and I ended living at Ronald McDonald House in NYC for more than 2 years. Today, I want to share this infinite gratitude that my family and I have with CRN, a community that feels like family, they gave me emotional and financial support to put the keystone in place and helped me to rebuild and be self-sufficient for my family. Thank you, thank you so much for helping me and giving me this life-change opportunity.

– HERBERTH SIGLER

”



Global Genes Rare Patient Advocacy Summit

By Jonathan Dicks



The 2021 Virtual Global Genes Rare Patient Advocacy Summit started as a blur as my alarm snapped me awake at 4:30am on a cozy Sunday. Why so early on a Sunday you say?

This particular Sunday in late September would prove to be my perfect introduction into rare patient advocacy from an international perspective. Advocacy leaders from Australia, New Zealand, Malaysia, Singapore, and Thailand recounted their unique patient advocacy stories that gave incredible insight and perspective in traversing this regional rare disease landscape. The next morning leaders from South Asia discussed their individual regional advocacy efforts, while advocating for better disease awareness campaigns, as well as stakeholder education, and increasing access to diagnosis, care, and disease management. I found it inspiring how so many miles separate us, yet our stories seem so familiar, almost like family.

We ended the night with RARE Together, Watch Together: an amazing sampling of nine short documentaries on the rare patient experience. From very young children to adults and even a woman who wasn't diagnosed rare until age 64. These incredibly

beautiful stories show us all how to be courageous when we are afraid, to have hope when others do not, and what the power of a focused community can accomplish in the face of seemingly insurmountable odds.

Fyodor Urnov, PhD gave a wonderful presentation highlighting the advancements in gene-based therapies such as gene-based diagnostics, therapies, and platforms like gene editing which are providing hope for faster progress towards cures.

I heard a terrific presentation on "building your own coping toolkit" as parents and caregivers. It reminded me of the heaviness and disbelief we

felt when diagnosis finally came, but the speed at which we had to catch up saw us running miles a minute, getting medications, blood draws, appointments, repeat. The journey to rock-solid advocacy is overwhelming and exhausting, but oh so worth the fight and can be done in a healthy way both mentally and physically.

Global Genes and the 2021 Rare Disease Patient Advocacy Summit did not disappoint and continues to be a shining example of what we can do as a rare community. I was honored to attend and encourage us all to re-align ourselves with the firm belief that our voices are beautiful and our stories inspire hope.



Family Support Committee Update

By Jen Wyman, Vice President of Family Support

There are a lot of articles out there that advise us to “live in the moment”, to focus on the present and what is right in front of us. But is that good advice? When dealing with a rare disease sometimes the standard advice doesn’t work or make sense.

“Our memories of experiences are more important than the experiences themselves. Before we know it, the day is over. The year is over. The kids are grown...but we have pictures and memories of these moments that last a lifetime and forge our relationships.” Time is fleeting. “When you live in your past, you will consistently make better, often harder decisions.”

On the flip side “living in the present moment means letting go of the past and not waiting for the future. It means living your life consciously, aware that each moment you breathe is a gift.”

And then there is the idea of not wishing your life away and living for the future. But looking to the future “shifts our attention from the monotony of the mundane to something more meaningful. The future becomes an emotional target that gives us purpose and helps us manage our frustration.”

It’s all very confusing. It is my experience that there are times in our lives where each of them is appropriate and right and downright necessary in the rare disease world.

Cystinosis has a way of making us live in the moment, focus on the past, and long for the future all in the same day. It’s what gets us through the day, the month, the year, our lives.

Our daughter, Kacy, was recently hospitalized with COVID. She was vaccinated and boosted and she was off to college. The world was



her oyster. THEN...three days into her freshman year she was sick, two weeks later she was hospitalized, and two weeks after that she was discharged. It was touch and go for a few of those days, so you can believe that we lived each of those past, present, future moments during that time. It is what kept us sane. Of course we longed for the past... that month before when we were getting her ready. It was an exciting time. Remembering that experience and knowing it was a possibility again was a good thing. But those two weeks in the hospital (as you ALL well know) are when we live in the present. It’s where we focus on the next lab draw, the next test, the next doctor visit. It’s hard to see outside of those moments and yet it’s necessary to get us to the next moment. I can tell you during

those moments I am NOT sitting back “grateful for each breath”. I’m just surviving. Hospital time is weird time. It moves fast and stands still all at the same time. And while these present moments are taking place we have glimpses of the future....longing for the future, when this will be behind us and things will go back to “normal” and she’ll be off to college again.

So it’s possible...to live in the moment, long for the past, learn from the past, look to the future and wish for the future all in the same day. It’s called survival and it’s sometimes what gets us through the bad days. Sometimes the goal is to just get through the day...or it’s important to not just get through the day, but look to the future-because that is where hope lives.

Oscar's Story

By Jenna McKenzie



Oscar was born in January 2020. He was a very big baby at 75cms long; 4.54kgs in weight; and with a massive 38cm head. He was solid from the get go and loved his food. He was such a hungry little fella that we were referred to a pediatrician at three months old because his size was tracking at the very top end of the scale and we were advised to start him on solids early. Things were progressing well and he was a very happy and contented baby. It was only when Oscar was ten months old that things began to fall apart. Oscar began vomiting in the night after sculling down water. We thought it was teething as his cheeks would go red and he was very hot to the touch. He would want water all the time and would get upset when we wouldn't offer another bottle after he had smashed down the first bottle. We went to our local GP to query the amount of water he was drinking. We were told that it was normal and he just liked his water. We queried the vomiting but were told it was likely

reflux and not to worry as all babies had some form of reflux.

We first noticed that Oscar was trimming off at around 10.5 months old. Oscar had begun walking super early at nine months, so we put the weight loss down to him being really active.

It was only over Christmas when he was standing in his swimmers that we saw how much weight he had lost and his legs had bowed out. We knew something wasn't right and we booked in to see the GP at their next available appointment.

We didn't make it to that appointment as the day after his first birthday party everything went downhill fast. We were out with my parents for breakfast, something Oscar loved to do but after inhaling the food he had a massive projectile vomit. We took him home and he seemed to perk up again so we put it down to him having caught a 24-hour virus from daycare. The very next day he deteriorated further including falling asleep on the

change table. We raced Oscar to the children's emergency department where he was weighed at only 8.7kgs - much to our absolute shock and horror. We couldn't work out how our little boy had deteriorated so quickly from a healthy 9.9kg baby at six months and to now being diagnosed with a failure to thrive. We were very distraught and blamed ourselves that we were not feeding him correctly. We questioned everything that we had been doing.

He was quickly admitted to ED for the failure to thrive and they found that he also had tonsillitis. An urgent appointment was made with a pediatrician. We were nearly discharged at this point, however, his bloods were taken and they saw that his potassium had dropped to 2.5 and that he was in severe hypokalemia. It was all systems go from here and as it was during COVID; Tim had to quickly race back into the hospital. Oscar continued to deteriorate that night and a nasal gastric tube was

inserted along with what felt like a million cannulas. At this stage, the ED registrar thought it was possibly renal tubular acidosis. A nephrologist specialist happened to be working in ED that night and popped in and saw Oscar. It was mentioned to her that Oscar may have a kidney condition, however it wasn't confirmed yet. We spent the night in ED waiting for a bed in the Paediatric Intensive Care Unit (PICU). The next morning, Oscar was taken to have his wrist x-rayed for rickets and we were rushed up to PICU as a bed was finally available.



We spent seven days in PICU which was truly the most devastating and scary moment in our lives. We were so very close to losing Oscar as his levels of critical minerals were unable to be stabilized. His life was literally in the hands of the PICU team. He was constantly vomiting, not being able to hold down any of the fluids or minerals in that were being pumped into him. The cannulas stopped working and we had to make the call to have a port put in his thigh. It was heartbreaking having to put your child through this and watch him being sedated by ketamine; all the while not knowing what was going on. But this



port was the thing that saved his life and allowed the PICU team to finally be able to treat him by being able to monitor his bloods continually and make the critical decisions as to what was needed to get him stable.

It was also during this time that the specialist nephrologist who saw Oscar in ED, was called into PICU to make the call as to whether Oscar was dealing with a genetic kidney condition. It was incredible timing as there was also a genetic trial available and we were able to have our bloods taken to confirm whether Oscar held the gene mutation for the genetic condition with results available in 2-3 days. We still had no idea what Oscar had only that it related to his kidneys and that it was genetic with myself and Tim being the carriers.

After a week and a half in PICU, Oscars electrolyte levels had started to stabilize and we were moved into the ward. We spent another week and a half in the hospital where he continued to improve. Oscar had to re-learn to walk and his strength throughout all of this was just amazing to witness. It was here that we finally met with the specialist nephrology team and they informed us that Oscar had Fanconi Syndrome and Nephropathic Cystinosis. We didn't realize it at the time however that we were lucky to have had such a quick



diagnosis and now looking back we are extremely thankful for this as we know a lot of other families went through long ordeals with a lot of misdiagnoses.

We had looked up earlier that Oscar may possibly have Fanconi Syndrome but missed the part where it said that Fanconi Syndrome is often the first sign of cystinosis. It hit us hard when we realized what Oscar had and what it meant for his life. We were devastated – for the life our little boy was meant to have. Having to consider your own child's mortality was utterly heartbreaking and it broke us, that moment was life changing and we as parents are not the same people we were before that diagnosis. It stays with you forever.

We were told about what medications Oscar would require on an ongoing basis, which was extremely overwhelming. Being told that you must administer Cystagon every six hours along with electrolytes - Phosphate, Magnesium, Potassium and Sodium Bicarbonate every eight hours whilst working out which ones can go together and which ones can't as it will make him vomit and somehow manage his nutritional needs by administering his feeds via his nasal gastric tube by a machine pump four times a day, was a really harrowing experience. It really hit



home just how quickly our life had been turned upside down. You question how you are going to get through it, knowing that you are now going to have to manage your son's endocrine system by balancing his electrolytes that are not only essential for basic life functioning but also extremely critical because an imbalance can have serious consequences. It's a lot to wrap your head around and we spent a lot of time in the ward learning as much as possible so we would be prepared and ready for when we went home.

We got home from the ward three days later and it really hit home just what this diagnosis meant moving forward.

We had to buy an IV pole for the lounge room where Oscar's feeds are administered, vomit bags and pee pads for his cot as he has polyuria. Hydrolyte was bought in bulk for vomiting. We moved a mattress into Oscar's room so one of us can do the night shift which involves changing nappies and doing his medications at 1am.

Our kitchen now has a medication trolley where we make up all his

medications for the day. We have hospital bags ready to go at any time as a simple gastric upset can send him straight into hospital.

A cupboard in the kitchen is now full of all of his enteral fed meals (tube fed). He has four pump feeds a day to support his nutrition and growth and the only way to have him sit still is watching tv. We have watched a lot of TV this year.

Weekends are spent going to allied health appointments to make sure we are supporting Oscar as much as possible and we have a whole team of professionals providing much needed support from orthotic shoes to support his legs from the rickets to an endocrinologist to support his growth and a speech therapist as his speech is quite delayed. There will be more added to the team as he gets older.

It's been a huge year full of grieving, acceptance and growing. Now looking back, we can say we have gotten through the worst of it for now and are in a place of wanting to give our beautiful boy the most wonderful life possible with the support from both of our families who have been incredible during this time.

We also wouldn't have been able to get through the first year without the amazing support from the families who have also gone through a diagnosis of cystinosis, the parents who have walked this road many times before. They have been a wealth of knowledge and it has been so comforting just knowing that we can reach out to a parent late at night after Oscar has vomited up his Cystagon for ideas on how to limit this happening in future. We are so grateful to have the parents group and we lean on it for so much.

We are looking forward to seeing the end of the year in a much more positive light and hold out hope for the stem cell therapy becoming available for all of those suffering from cystinosis.

Professional photos courtesy of Fiona Lauren Photography.



Open Letter of Gratitude

By Asim Bukhari

There is a saying in our language that when all the doors are closed in times of trouble and need, Allah opens for you the one door where He wants to take you and in that there is a blessing in it.

Face the difficulties ~ because there is no difficulty in your life that no one else has faced. There is no tragedy that does not happen to anyone else. And of course, there is no success that no one else has. We have never been introduced to a rare disease, and it is a disease, that is still unknown to many in our society.

Under these circumstances, our connection to the Cystinosis Research Network was no less than a blessing. Every disaster brings an opportunity. Covid-19 will never be remembered as acceptable trouble, but it did give us

an opportunity to be part of the annual virtual conference that would have been very difficult for us under normal circumstances. The conference was very well organized. We had a great opportunity to get information about cystinosis from many medical experts, and at the same time, we could talk to them about our problem. It was very important for me to get as much information as possible about this disease and that is why I was interested in taking sessions with the medical panel and parents.

Talking to people all over the world was very useful and as a result of that, the fear and delusion that was in the heart about this disease are definitely no more. We got acquainted with a lot of people and it happened because of the online conference and because of the belief that Allah will surely guide

us in our trouble. It is very important to hold the in-person conference because in virtual events you have to give everyone equal time but the biggest advantage of a virtual event is that people from all over the world can benefit from it at the same time which might be limited to a few in physical presence.

Overall, it was an excellent event but if you allow me to add a topic then I would include a motivational speaker. This might add value since motivation is everything that keeps hope alive and many families might need that.

Last but not least, on behalf of every person, thank you again for organizing this wonderful event.

Asim Bukhari (Father of a 6 year old son with cystinosis)

Saudi Arabia



Cystinosis Memorial Fund

By Karen Gledhill

The Cystinosis Memorial Fund (CMF) was established to support teens and adults living with cystinosis, while honoring the warriors who have lost their battle. Our goal is to instill added confidence and improve the skills and abilities of recipients in our community to flourish and fulfill their potential.

The CMF will provide a stipend to those living with cystinosis of up to \$1,000 USD. Professional workshops and certifications, career and resume coaching, purchasing new software programs, and tutoring are just a sample of the professional and technology-focused goods and services covered.

This year, seven of our community members received awards ranging from \$400 to \$1000. Congratulations to Niall Barron, Kellen Binger, Melina Castro, Hannah Creel, Emily Patterson, Cheryl Simoens, and Tahníe Woodward.

Considering applying? Applications are being accepted at <https://cystinosis.org/cmf>. Here's feedback from one of our first CMF award recipients, Niall B:



“

You should apply to the Cystinosis Memorial Fund to show children with cystinosis what's possible with a bit of effort. You're constantly reminded as a child of what you won't be able to do because of cystinosis, to the point that you dismiss a lot of ordinary life experiences. Showing newly diagnosed kids some examples of accomplished and successful adults with cystinosis will encourage them to challenge themselves to set their goals a little higher.

CMF also makes the application process as simple and straightforward as possible, so applying takes very little time. ”

– NIALL B.

PROCYSBI Update from Horizon

Starting in November, PROCYSBI PACKETS will begin to arrive in 60 count cartons, instead of 120 count. This does not change the price you pay, nor does it change your dosage. For some patients, this change may not occur for the next few months, it all depends on the timing of when shipments occur. Horizon is making this change to ensure efficient

delivery of your medication. If you are receiving a 120 count carton you will now receive two 60 count cartons. Nothing else will change with your PROCYSBI PACKETS. Please contact your Patient Access Liaison (PAL) or email connect@horizontherapeutics.com with any additional questions.



Cystinosis Research Network - Board Member Updates

Over the summer, the nominating committee proposed a new slate of board and executive committee members for open positions and a vote was held. As a result, we are proud to announce the following:



Jonathan Dicks – President

Shortly after his daughter’s cystinosis diagnosis, Jon knew he couldn’t sit on the sidelines and let this disease run the show. He immediately began advocating for his daughter, Eleanor (Elle), and has not looked back. For the past few years, Jon has held the position of CRN VP of Development. He took the responsibility to heart and dove in head first, proving he was a dedicated asset to our community. Please help us in welcoming Jon into his new position as President.



Tim Wyman – Treasurer

Professionally, Tim is the Managing Partner and Certified Financial Planner professional at Center for Financial Planning, Inc. He has received national recognition including the Forbes’ Best-In-State Wealth Advisors List in Michigan, as well as, Financial Times 400 Top Financial Advisor. This skill set will make for a natural transition from CRN Interim President to Treasurer. Personally, Tim and his wife, Jen, have three children. Their youngest child, Kacy, was diagnosed with cystinosis in 2005, had a successful kidney transplant in 2015 and started at Grand Valley State University this fall.



Karen Gledhill – Secretary

Karen is one of the founding members of the Adult Leadership Advisory Board (ALAB), a group comprised entirely of adults living with cystinosis. Karen’s willingness to tackle challenging topics with empathy and sensitivity makes her an important voice to have in the room. We look forward to including her perspective on the Board during her term as Secretary.



Kristina Sevel – Director

Kristina rounds out our newest CRN slate as a Director. Relatively new to the cystinosis community, her daughter (Grace) was diagnosed in 2020. Shortly after, the Sevels connected with the CRN and held their first fundraiser. During a whirlwind year, Kristina and her husband, Mike, shared some of their story via The Cystinosis Advocate newsletter. You can learn more about the Sevels and “meet” sweet Grace in the Spring/ Summer 2020 newsletter at cystinosis.org/our-newsletter.

We'd like to recognize outgoing Secretary, Ina Gardener, and outgoing Treasurer, Jenni Sexstone, for their service and countless volunteer hours. Thank you!

CRN Board of Directors

- Jonathan Dicks, President and Vice President of Development
- Christy Greeley, Vice President of Research and Executive Director
- Jen Wyman, Vice President of Family Support
- Marybeth Krummenacker, Vice President of Education and Awareness
- Tim Wyman, Treasurer
- Karen Gledhill, Secretary
- Carol Hughes, Director
- Gail Potts, Director
- Kristina Sevel, Director
- Herberth Sigler, Director
- Sara Healy, Adult Patient Representative



New CRN Board Member Perspective

By Kristina Sevel

My name is Kristina Sevel, I am honored to be one of the new board members for CRN. My husband Mike, daughter Grace and I live in the Cleveland, Ohio area. Grace is now 2.5 years old, and was diagnosed with cystinosis in May of 2020. Outside of being a mom to Grace, I work as a RN Care Coordinator for the Cleveland Clinic. Prior to cystinosis I had worked for four years in ophthalmology. After navigating the challenges of being on the patient side of things I knew

that with this new knowledge I could use some of our experiences to help others during and after a hospital stay. Our daughter was diagnosed with cystinosis in May of 2020 during the initial peak of COVID. We spent a total of 20 days in the hospital getting Grace well and along with all of the new medications and routines came isolation and loneliness. Not only were we dealing with an ultra-rare disease, but with COVID visitation in place we weren't able to have any of our close

family or friends around to support us. We joined the groups, got in touch with other parents who offered to call and reassure us that we were going to get through this, but it was Jen (Wyman) and other CRN board members and parents who helped pull us out of the hole that we felt stuck in. My goal is to help in passing on the support that we received when the world felt like it had crumbled around us in those early days.

HAVE YOU OR YOUR CHILD BEEN DIAGNOSED WITH CYSTINOSIS?

you/they may qualify for a research study looking at how the brain works in cystinosis

THIS IS A 3-DAY STUDY INCLUDING EEG AND COGNITIVE TESTING.

TRAVEL AND ACCOMMODATION EXPENSES WILL BE COVERED



CONTACT

**ANA.ALVESFRANCISCO@EINSTEINMED.ORG
OR 718-862-1824 TO KNOW MORE!**



Development Committee Update

By Jonathan Dicks, Vice President of Development

We are in full swing with the holiday season fast approaching which means in my role as VP Development proposals for the 2022 fiscal year are hot off the printer. I expect to be humbled yet again by the generosity of our industry partners, 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. Last year we were able to secure over \$280,000 in direct funding, and I know next year will be even more impressive, as our growth dictates increased needs.



Fighting the good fight; Elle, Shirley and Finn Dicks.



pcsforpeople

PCs for People (if you haven't heard) can help you as a patient or caregiver in need of a new computer, and at absolutely zero cost. Send me a message at jdicks@cystinosis.org and I'll reply with a brief questionnaire and get the entire process facilitated from start to finish. It really is that simple.

If you tuned in for the 2021 CRN Virtual Family Conference this past summer and were surprised to see Spanish interpreters in every breakout session, please thank Horizon Therapeutics and their partnership with Bromberg & Associates who offer the CRN translation and telephonic interpretation for Spanish-speaking

individuals and families impacted by cystinosis.

Our friends over at the American Kidney Fund have launched a digital fact sheet to help you successfully prepare for and navigate through a telehealth physician visit. Check it out on page 23!

CRN and the Center for Chronic Illness are still offering free, professionally facilitated support groups and health education programs for those impacted by cystinosis as well as a separate monthly group specifically for parents with a diagnosed child.

Holidays are a time of giving so please keep checking in on all our social media accounts for some awesome events all over the USA and abroad. With only a few months left in 2021, there's still plenty of time to make a great personal impact into local outreach and advocacy events.

Don't see one? Give us a ring and we'll assist in mapping a plan of action. Don't forget, 2022 Rare Disease Day is quickly approaching on February 28th and Cystinosis Awareness Day on May 7th.

The fundraising side had a wildly successful turnout for the 1st Annual Cystinosis Research Network Charity Golf Tournament on August 14th, raising over \$7,000 in just an afternoon of golf (see page 22 for details). Incredible advocacy work Gail, we are so appreciative of your dedication, calm wisdom, and grace. We can't wait to watch this event grow year after year!

Have a question about how to do something like this? Or maybe you have a unique fundraising idea but are unsure of where to start? Be on the lookout for a "fundraising playbook" coming in 2022 for quick and easy tips on successfully navigating the fundraising area, as well as a how-to guide for starting your own 501(c)(3).

CRN Myrtle Beach Golf Outing

By Gail Potts



The CRN golf tournament held in Myrtle Beach South Carolina at Myrtle Beach National Golf Course was able to raise over \$7,000. The event was held on a hot blustery day, August 14, with 96 golfers participating. The morning started off with registration and a breakfast sandwich from Bojangles. Golfers were able to purchase mulligans and raffle tickets at registration. Area merchants donated many of the prizes, some of which included restaurant dinners, massages, spa treatments, golf accessories and more. We were fortunate to have Jeff Larimore, a past CRN President play and BJ Viau from Horizon Therapeutics participated. Jeff was kind enough to offer a few words to our golfers about CRN at the start of the tournament. Dixon Golf was present to offer prizes for golfers participating in their challenges, such as, closest to the pin. Players paid \$10 or \$20 to participate in the challenge. They donated a portion of the money collected for this event to CRN as well.

Besides raising funds for CRN, I was able to interact with friends and visitors in sharing information about cystinosis and raising awareness. I

started out on this journey with no prior experience in fundraising and having to raise awareness about a rare disease of which no one had heard of. I did a lot of praying and my prayers were answered. My thanks go to God for the success he provided for this tournament by having friends step up to listen to my cause, help by spreading the word and participating in the tournament. Some friends volunteered. Others played in the tournament. Some helped acquire raffle prizes. Others helped put together baskets for the raffle prizes. I also had someone help with media visibility.

I had the privilege of meeting Kery and Tom Westfall, Tori and Addyson Westfall prior to the tournament when they reached out to me after seeing the flier on the CRN website. Addyson is two years old and was just diagnosed in May of this year. You can learn more about Tori and Addyson's journey in The Cystinosis Advocate Spring/Summer 2021 edition.

It was awesome having Kerry and Tom Westfall volunteer for the event. While we waited for the golfers to finish their round of golf and return for lunch, we had a chance to talk

and meet some visitors who showed an interest in learning more about cystinosis. This was a real plus for the tournament as it gave us a chance to educate the public and raise awareness. Thank you so much for volunteering and offering information about cystinosis to our visitors interested in learning about it.

Besides raising money for CRN, it was successful in bringing awareness and uniting two families with children with cystinosis. I would call it a successful event.





Virtual doctor visits and cystinosis



When you have cystinosis, it is important to keep up with your doctor visits and treatments. This helps you slow damage to your kidneys and other parts of your body and stay healthy. Many doctor's offices now offer virtual visits (also called telemedicine or telehealth visits).

Virtual visits let you see your doctor from home – or wherever you are.

You may have a **virtual visit** between in-person visits, or in place of an in-person visit. In a virtual visit, you often use an app or online patient portal to connect with a doctor in real time from a distance.

Here is how to get the most out of your virtual visits:

1. Prepare for your virtual visit

Make sure your device is ready

Use a smartphone, tablet or computer with a webcam for your visit.

- Choose the device that has the best camera
- Test the camera and sound
- Make sure you have a good internet connection

Everyone doctor's office is different. Your doctor's office sends you a specific app or link to use. **Make sure you download the app or click the link before your visit to make sure it works.**

Right before your visit starts:

- Fully charge your device or plug it in so you do not lose connection
- Close other apps or programs

Set up the room for your visit

- Find a quiet, private place with little background noise – you may need to share personal details with your doctor that you would not want others to hear
- Make sure the room has plenty of light
- Sit eye-level with the camera – this helps you make eye contact with your doctor
- Keep your device steady during your visit, such as by propping it up on a table
- Make sure you are comfortable with your doctor seeing what's in your background

Gather what you may need

You may need:

- All your medicines, or a list of them, to review with your doctor
- Thermometer
- Flashlight – your doctor may ask you to shine a light to see a part of your body close up
- Blood pressure monitor
- Scale to check your weight
- Blood glucose meter, if you have diabetes



Wear loose, comfortable clothing – it will be easy to move if you need to show your doctor a certain part of your body

Write down a list of your questions before the visit starts

Here are some to start:

- What can I do to care for my symptoms?
- What should I do if my symptoms continue or get worse?
- What do my most recent lab results mean?

Tip: Make sure to get your lab work before your telehealth visit.

If you are preparing for a kidney transplant, you may want to ask:

- How long will the surgery last?
- How soon can I go home from the hospital after surgery?
- Can I have medicine if I feel pain?

2. Ask questions and get answers

Describe your symptoms to help your doctor give you the right care.

For example, tell them:

My symptoms include (check all that apply):

- Trouble seeing, watery eyes or sensitivity to light (your eyes react by closing or squinting when you see a bright light)
- Trouble swallowing
- Muscle weakness
- Feeling very thirsty
- Peeing more than usual
- Other: _____

My symptoms have lasted _____ (length of time)



Ask your most important questions first in case you run out of time.

Check that you understand what your doctor said by repeating the information back to them in your own words. You can say:

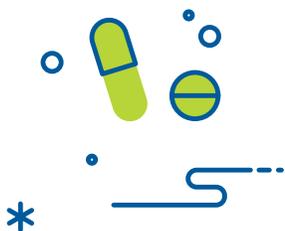


"I think what you are saying is... (repeat the information). Is that right?"



Ways to manage cystinosis between virtual visits

Focus on taking your medicine and caring for your health so you can keep doing all the other things you like!



To manage cystinosis :

- Keep taking your regular medicines for cystinosis as prescribed at the same time every day and stay on all medicines that your doctor prescribes for you to take after your transplant
- If you had a kidney transplant, take your immunosuppressant medicines as prescribed, too – they protect your new kidney from being attacked by your immune system
- Follow a healthy meal plan, including supplements if your doctor recommended them
- Be active most days of the week – pick activities you enjoy, such as walking, dancing or playing a sport
- Reach out to your family and friends when you need to talk – it is normal to have many feelings, such as feeling anxious, stressed or unhappy. You do not have to deal with your feelings alone.
- If you would rather talk to someone you do not know, ask your parents or doctor to help you find a counselor. You can also reach out to the Cystinosis Research Network (cystinosis.org), who can connect you to support groups and many other resources.

Learn more about cystinosis and kidney disease at kidneyfund.org/cystinosis

Live like Laura Fun Fund Update

By Frankie McGinnis

I am pleased to say that we have been able to supply six grants with a 7th and 8th award in process. What started as a way to keep the love of life that my Laura had spreading, has become such a blessing to me on my darkest days. Who knew the people that would apply would send applications in and I would receive them at a time when the waves of grief were overwhelming me? The knowledge that Laura's memory lives on, that I am able to spread love and laughter in her name is such an honor. I am happy to share some stories of the people who received grants. May you live your life fully and with joy.

Peytan's Grant

By Channing Taylor



Peytan, 11 years old, was diagnosed with cystinosis at 15 months old. Many doctor appointments and tests led us to the final diagnosis. She has spent most of her life in hospitals for numerous issues.

Peytan suffers from anxiety and PTSD from some events that happened a few years ago where she thought she wouldn't pull through. Recently she has found a love of horses. We call her the animal whisperer as she has always had a way with animals and them loving her.

A dear friend posted on Facebook about working with horses and giving riding lessons. I thought it was the perfect opportunity to help Peytan with her anxieties. I reached out to her and shortly after we started having Peytan go out for therapy sessions there. She instantly fell in love with all of the horses and a special donkey.

I had followed Laura's story and met Frankie a few times through some meetups for cystinosis. So when I saw that to honor Laura she started the Live Like Laura grant I thought, why not try to apply so Peytan could continue her therapy sessions?

To my surprise we were chosen for the grant! Peytan was able to get about four months out of the grant of weekly therapy sessions! We are so thankful for the opportunity and hope that Peytan Lived Like Laura and made her proud.

Ashley Abedini

Chronic Illness, Mental Illness, and Entrepreneurship. These are the things I have been exploring with my podcast, The Bipolar Nextdoor. I recently spent some time in graduate school for my LPC or counselors degree. After tackling my own diagnosis of Bipolar, I had a new found passion to help others. However, it didn't long for my overly empathic self to realize that I was not meant to be in a clinical position. Although I am not a counselor, I hope to help destigmatize mental illness by talking about it in my podcast.

We so often focus on the weaknesses of having a mental or chronic illness, however what if we started to utilize them as our entrepreneurial superpower? There is a huge overlap of business owners and those who have mental illness, the most notable being bipolar and ADHD. The very traits that describe a bipolar person are the same traits that



make a good business owner often. I have had a great time talking to local business owners in my town, Wichita. I have been able to interview successful women business owners, with diagnoses ranging from bipolar and ADHD, to depression. I want to not just expose others to mental illness diagnoses, but to show successful examples of utilizing it for their professional goals. I plan to further discuss chronic illness later in this season. There is a huge correlation with chronic illness and mental illness that is undeniable.

I am extremely grateful for the Live Like Laura Fund. I did not know Laura well, however, I felt like I had a true sense of her ambitious spirit listening to everyone's beautiful descriptions of her vibrant life. I remember feeling like I needed to live life a bit more intensely reading her obituary. That's an advantage of having cystinosis, there's a sense of urgency that is self aware. You know more than anyone that life is fragile, and it tends to put your goals at the forefront. I am grateful to be a recipient of the Live Like Laura Fund. I plan to officially launch the podcast next week. I have been using the money received to record and film. This has allowed me to dive deep into an interest I've had for a while.



The Adult Leadership Advisory Board (ALAB) is a group comprised entirely of adults living with cystinosis. ALAB's mission is to share stories and strength to educate, motivate and empower the entire cystinosis community. Through partnerships with the CRN and other organizations, ALAB focuses on issues and challenges through developing programs, opportunities and mentorship. Learn more at cystinosis.org/ALAB.

Update: Adult Leadership Advisory Board

By Sara Healy

Hi Cystinosis Community,

Our next podcast episode for Cystinosis Rare: Journey Into the Unknown is about coping mechanisms developed when living with a rare disease or taking care of someone with a rare disease. We are looking for siblings and adults with cystinosis to participate in our podcast to tell their point of view on this subject. Please reach out to us by December by private message on our ALAB (Adult Leadership Advisory Board) Facebook page or email us at alab47213@gmail.com. We will be doing a two-part episode; the first episode will be the adults with cystinosis and will air mid-January and the second will be cystinosis siblings at the end of January.



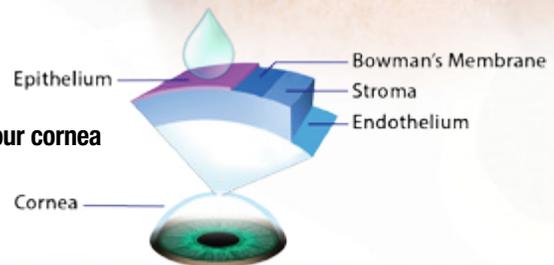
Our Adult Leadership Advisory Board wants to know how you think we are doing as a group. We would like your feedback on what you like about our programs and how you think we can better help the community. We are here to help our community thrive and make sure you feel heard. Please fill out our survey <https://www.surveymonkey.com/r/ALABsurvey> or email directly at alab47213@gmail.com with your feedback. We would love to hear from you.



Special drops for special eyes... ...your eyes*

- **CYSTADROPS** is a viscous solution that coats the cornea
- **CYSTADROPS** can dissolve cystine crystals throughout the layers of the cornea
- **CYSTADROPS** dosing schedule is 1 drop 4X a day

CYSTADROPS penetrates all layers of your cornea



Indications and Usage*

CYSTADROPS is a cystine-depleting agent indicated for the treatment of corneal cystine crystal deposits in adults and children with cystinosis.

Important Safety Information

- To minimize the risk of contamination, do not touch the dropper tip to any surface. Keep bottle tightly closed when not in use.
- A condition where the pressure inside the skull increases for unknown reasons has been reported with cysteamine taken by mouth or cysteamine eye drops (used at the same time as cysteamine taken by mouth). This condition went away with the addition of medicine that increases the production of urine.
- Contains the preservative benzalkonium chloride. Contact with soft contact lenses should be avoided. Remove contact lenses prior to application. Lenses may be reinserted 15 minutes following administration.
- The most common side effects are eye pain (stinging), blurred vision, eye irritation (burning), eye redness, discomfort at instillation site (sticky eyes or sticky eyelids), eye itching, watery eyes, medicine deposit on the eye lashes or around the eyes
- **To report SUSPECTED SIDE EFFECTS, contact Recordati Rare Diseases Inc. at 1-888-575-8344, or FDA at 1 800-FDA-1088 or www.fda.gov/medwatch.**

* For use by individuals with cystinosis

Visit www.cystadrops.com for full prescribing information



PATIENT BRIEF SUMMARY

CYSTADROPS® (sys-tah-drops) (cysteamine ophthalmic solution) 0.37%, for topical ophthalmic use

Summary:

Read this information before you start using Cystadrops and each time you get a refill. There may be new information. This information does not take the place of talking to your doctor about your medical condition or your treatment.

What is Cystadrops?

Cystadrops is a prescription cystine-depleting solution used for the treatment of corneal cystine crystal deposits in adults and children with cystinosis.

What should I know about using Cystadrops?

- To minimize the risk of contamination, do not touch the dropper tip to any surface. Keep bottle tightly closed when not in use.
- Cystadrops contains the preservative benzalkonium chloride. Contact with soft contact lenses should be avoided. Remove contact lenses prior to application. Lenses may be reinserted 15 minutes following administration.

Before you use Cystadrops, tell your doctor if you:

- are using any other eye drops
- wear contact lenses
- are pregnant or plan to become pregnant. It is not known if Cystadrops will harm your unborn baby.
- are breastfeeding or plan to breastfeed. It is not known if Cystadrops passes into your breast milk. Talk to your doctor about the best way to feed your baby if you use Cystadrops.

How should I use Cystadrops?

See the complete Instructions for Use for detailed instructions about the right way to use Cystadrops.

- Use Cystadrops as your doctor tells you.
- Use one drop of Cystadrops in each eye, four times each day.
- If you miss a dose, administer the dose as soon as feasible, and then continue the treatment with the next scheduled dose.
- Keep Cystadrops in the refrigerator until ready to use. After opening, write date on carton and after each dose, keep the bottle tightly closed and store at room temperature in the original carton.
- Discard bottle at the end of 7 days, even if medication is still in the bottle. The solution is only stable for 7 days after opening.

What are the possible side effects of Cystadrops?

The most common side effects are eye pain (stinging), blurred vision, eye irritation (burning), eye redness, discomfort at instillation site (sticky eyes or sticky eyelids), eye itching, watery eyes, medicine deposit on the eye lashes or around the eyes.

A condition where the pressure inside the skull increases for unknown reasons has been reported with cysteamine taken by mouth or cysteamine eye drops (used at the same time as cysteamine taken by mouth). This condition went away with the addition of medicine that increases the production of urine.

These are not all the possible side effects of Cystadrops. Tell your doctor if you have any side effects that bother you. You may also report side effects to **FDA at 1-800-FDA-1088**.

How should I store Cystadrops?

- **Before First Opening:** Before opening, store new, unopened CYSTADROPS in the refrigerator between 36°F to 46°F (2°C to 8°C). Keep the bottle in the outer carton in order to protect from light.
- **After First Opening:** After opening, store opened CYSTADROPS at room temperature between 68°F to 77°F (20°C to 25°C). Do not refrigerate after opening. Keep the dropper bottle tightly closed in the outer carton in order to protect from light. Discard 7 days after first opening.

Keep Cystadrops and all medicines out of the reach of children.

General information about the safe and effective use of Cystadrops.

Medicines are sometimes prescribed for purposes other than those listed in a Patient Information leaflet. You can ask your pharmacist or doctor for information about Cystadrops that is written for health professionals. Do not use Cystadrops for a condition for which it was not prescribed. Do not give Cystadrops to other people, even if they have the same symptoms you have. It may harm them.

What are the ingredients in Cystadrops?

Active ingredient: cysteamine 3.8 mg/mL (equivalent to cysteamine hydrochloride 5.6 mg/mL);

Preservative: benzalkonium chloride 0.1 mg/mL;

Inactive Ingredients: carmellose sodium, citric acid monohydrate, disodium edetate dihydrate, hydrochloric acid and sodium hydroxide (to adjust pH to 4.6-5.4), and water for injection.

Manufactured by: Baccinex SA, 2822 Courroux, Switzerland

Manufactured for: Recordati Rare Diseases Inc., Lebanon, NJ 08833, U.S.A

For the most recent prescribing information, please visit

www.recordatirarediseases.com/us

Revised: June 2020

Providing support every step of the way for patients taking Cystadrops



Scan QR Code
to enroll now

- Working with your insurance company to confirm coverage
- Providing Copay Assistance Program for eligible patients
- Personalized one-on-one support is available for patients receiving treatment with CYSTADROPS and their caregivers. Enrollment is voluntary.

To find out more – visit www.cystadrops.com/resources-faq/



CYSTADROPS is a licensed trademark of Recordati Rare Diseases Inc.
© 2021 Recordati Rare Diseases Inc.
Recordati Rare Diseases Inc. • Lebanon, NJ 08833
www.recordatirarediseases.com/us PP-CDPS-US-0108

 **Cystadrops**
(cysteamine ophthalmic solution) 0.37%



How a Rare Disease Impacts a Family:

A Global Genes RAREcast interview with Jon and Shirley Dicks

Published October 8, 2021



Jon and Shirley Dicks' daughter Elle first started developing problems swallowing when she turned 10 months old. As her difficulty eating progressed, she stopped growing. Her parents grew frustrated by doctors dismissing her symptoms until a passing comment that she seemed to have a sensitivity to light allowed a specialist to diagnose her with the rare, lysosomal storage disorder cystinosis. We spoke to the Dicks about Elle's diagnostic odyssey, how having a diagnosis changed care for her, and the challenges they have faced caring for a child with a rare condition.

Daniel Levine: Jon, Shirley. Thanks for joining us.

Shirley Dicks: Thank you for having us.

Jon Dicks: Thanks, Danny.

Daniel Levine: We're going to talk about your daughter Elle, cystinosis, and what living with this condition has meant for you as a family. I'd like to start when Elle was around 10 months old and she began having problems swallowing. Shirley, what happened?

Shirley Dicks: Around 10 months old, Elle actually got a stomach bug and was sick for a few days. We noticed after she recovered from that stomach bug that when we presented table food to her, she didn't really swallow it. She just kind of chewed it up and spit it out or kind of played in it. And she would eat, she did eat, it was very

minimal amounts, like she would eat a little, a few bites of yogurt and a few bites of hummus, and that was about it. She really didn't swallow any food that was given to her.

Daniel Levine: It was after her 15-month appointment that things started to change. What happened then? When did you realize that there was something more serious going on?

Shirley Dicks: At her 15-month appointment, the doctor noticed that she had not grown any in height nor had she gained any weight in comparison to her 12-month appointment. She was not growing. She was not going up on the scale or on the curve. I did point out to the doctor that she still was having difficulty swallowing food and that she really wasn't interested in eating much. She was still only taking little bites of hummus and yogurt here and there and she was still spinning out a lot of her food. So, I did ask for a referral for therapy and I did get that referral. We started feeding therapy and we were also sent to a nutritionist who basically told us that we needed to get more calories in her to help her grow. And she just gave us a few recipes of different smoothies and things to try to get her to eat and get some more calories in her.

Daniel Levine: Jon, at the time you were working as an emergency department clinical care RN in a level one trauma center. As someone who works in a serious medical environment, what was going on in your mind? How good a handle on the situation did you think the doctors

had?

Jon Dicks: I mean, that's a very good question. I think that from the work, at that point in time I had been in the emergency department for about four years. And one of the things that we do really well in the emergency department is really know when to get worried. That was one of the hardest parts of this—that we were very, very worried and it seemed like we were the only ones that were that worried. It was, a very blasé attitude to what we felt like was a little girl that was kind of wasting away right in front of us, as evidenced by the plan of care that they had in place. I mean, they were kind of the elephant in the room that she didn't swallow anything other than breast milk, small amounts of food, and a ton of water.

So right off the jump, when they came back to us with, "Hey, here's this really, really thick smoothie and have her drink this," you just kind of cock your head, like that puppy look, and you're just like, "Did you not hear anything that we were talking about?" And it really made it clear that we were going to have to really try to steer where this was going to go because, and to be honest with you, Dan, at that point in time, we really didn't know where this was going, but we certainly knew that this was more than just an aversion of food or just a, a finicky or picky eater, uh, the way they kind of portrayed Elle to be.

Daniel Levine: Her health continued to worsen. There was a call you got from a resident in the emergency department of the local children's hospital. When did that happen? And

what happened with that call?

Shirley Dicks: We ended up getting that call because we had to follow up with our pediatrician at 18 months. And at 18 months she had actually lost some weight. So, she referred us to get blood work done through endocrinology. We went and got the blood work done. But prior to that, we also got sent to the feeding team at the hospital to check her out and see where things were going, and they also said it was neutral, so we just needed to get more calories and her. After that we went to see the endocrinologist. She did several tests, thyroid, diabetes, because we were saying how much she was drinking water, chugging it, wanting it all the time. She was sweating at night, like soaking through. So she got the blood work done. And I received a call while I was at work that day and the first thing the doctor said was, "Are you sitting down?" I said, no, but I can; I'm at work. And she said, "You're going to need to bring Elle in right away. Her labs are very concerning. She's very dehydrated and we need to get her back in and do some more blood work." She's like, "And you need the pocket bag to be ready to be admitted." So, I immediately left work right away or ran home to get Elle, got a hold of Jon, and we packed a bag and I went back to the hospital that day, got more lab work done. Same thing showed up—she was very dehydrated. They were very concerned about her labs; her electrolytes were pretty much bottomed out. So, we got admitted that night to the hospital and that night they placed an NG tube down her nose and throat immediately once we got into a room because they said she needed nutrition and she needed it quickly,

Daniel Levine: She's got this NG

tube. She's constantly pleading for water. The doctor at the time tells you that this constant demand for water is behavioral, which is kind of mind blowing to think. But at that point, Jon, you demanded she be discharged. Why and what happened?

Jon Dicks: Yeah, obviously it's a lived experience and I talked with Shirley and from my standpoint, we weren't really, the interventions that they were doing was really just to monitor. They really didn't have an understanding on why. Every time we asked them, "Why is she drinking so much water? And why is she so dehydrated?" you got answers like, well, sometimes we just don't know why kids don't want to eat or don't want to drink. So, I felt like if that was going to be the answer, then we can certainly do this in a place that's less intimidating, that has less beeping noises, that my daughter can actually get some sleep.

I think one of the benefits here is that I know how the hospital system works and I, and I kind of know how to push things a little bit. So, I made it very, very clear that we were not going to be spending any more time in here; that from what they were doing from our standpoint was something that we could do at home. As an emergency department nurse, an NG tube is not uncommon to see. You see people in the emergency department when they're in their worst state, right? We don't discriminate on who comes there. So, if somebody is having a very, very bad panic attack, but happens to have an NG tube, I'm expected to understand how to use that. So, none of that was really scary for us. And I felt like in the home setting, it would just be much better for Elle. So, we demanded the discharge and they honored that request with the stipulation that we follow up with the team of doctors to

which, of course, we were agreeable.

Daniel Levine: And how has her health progressing at this point? At what point did she progress to a gastric tube?

Shirley Dicks: So, she actually really struggled with the NG tube. Every time they pushed a formula threw it, or we pushed a formula into it, she threw up, she was constantly throwing up. She still wasn't gaining any weight. They were trying all sorts of different formulas, trying to push all sorts of different stuff to get her nutrition and nothing seemed to be working. She probably had that NG tube for, I think, eight months.

Jon Dicks: Yeah, she had it for eight months. I'll be honest with you, that was a very, very dark time for us because, you get the NG tube in and you say to yourself, "Okay, we've got this route to get nutrition in." But the elephant in the room from my standpoint was just that we still didn't understand what was causing this. So, we were being very, very reactionary with everything and that reaction wasn't working. The reaction to her not eating was let's put the NG tube then. And, you know, when I say like cyclical vomiting, in the medical terminology what that means is vomiting, you know, in excess of 10 times in a day, Elle was probably throwing up closer to 25 to 30 times a day, dry heaving, just could not keep anything down. As a father, the look of terror in your child's eyes when she's actually gagging back up the NG tube, and you're watching this tube come out of her nose and you've got to push it back in, you know, that's something that sits with me a lot, and unfortunately, we kind of dragged our feet on that to get her progressed to a G-tube. And I think it was just wanting to believe that everything was okay, and this was just a finicky little

eater, but the writing was on the wall, and we probably had that NG tube in probably about eight months, too long to be honest with you. In hindsight, and obviously hindsight being perfect vision, we should have just gone for the G-tube right then and there. I think had we had the diagnosis then we obviously would have gone to the G-tube. But it still haunts me to this day about the kind of lasting trauma that we put her through, the unnecessary trauma that we put her through there. I think that the 50,000-foot view is that we're doing all this, but we still don't have a real diagnosis and she's just continuing to get smaller.

Daniel Levine: And how old is Elle at this point?

Shirley Dicks: She's six years old right now.

Daniel Levine: No, I'm sorry. When she had the gastric tube for six months.

Shirley Dicks: Yeah. When did she get that tube? Jon, do you happen to remember? I can't even remember when she got it. She was two and a half, I believe. Yeah, because I remember she still had pictures on her second birthday of the NG tube.

Daniel Levine: You've been going through this ordeal for 18 months, two years at that point. What kind of a stress is that on you as care givers? Are you still trying to hold a job down at the same time or do you have any support from other family members?

Shirley Dicks: So, it was actually a very, very stressful time. I ended up going part-time because it was just so hard to get anything in her and she just kept losing weight, and it was a lot of commitment and responsibility to put on someone else to care for her, especially with the NG tube. So, my job was very understanding and I was

able to find a job share partner where I could end up working two to three days a week, and I actually still to this day continue to do that because she was needing so much out with the NG tube and we were having so many appointments and we were just really, really struggling to keep her healthy, and to figure out what was going on. So I did end up having to go part-time for that reason.

Jon Dicks: Yeah. And I kept full-time in the ER. I think there's probably a lot of reasons for that, obviously from a financial standpoint, we were quickly finding that our deductible went away like that. Right. So, at that first hospital stay, we had our deductible met for the entire year. Obviously, there's a financial strain that brings on the family and with that comes a lot of stress. For me personally, in the ER, I was finding it more and more difficult to give all of myself to these patients when I was crumbling on the inside myself. There was very, very little time as far as any time for Shirley just to be quiet with each other. That was just kind of non-existent, it was just you put out one fire and then the next one kind of pops up. It's one of those things where at least right now what we were understanding was that the minute we felt like we had one thing figured out that there was something else going on and it's all consuming. How do you bargain time with somebody that needs everything from you and is just pleading for someone to be an advocate for them? And so, our relationship definitely was rocky. I mean, there was a lot of times where we were just so off and angry and, you know, you try not to let any cynicism creep into it, but really we were dealing with people that at a point in time were kind of covering their eyes and throwing the spaghetti at the wall to see what would stick and they

have all these great ideas and these wonderful plans on paper, but then when you go home, the entire thing just blows up and it's something to be said in the rare disease community in the chronic disease community, you can't ignore how much that affects the family unit and the family life.

And I think we're doing so well right now it's only because we recognize that that was something that we had to really work at. Do we do a great job at it, sometimes. Right. But the reality of the situation was at that acute time, man, there were some very, very quiet terse moments. To be honest with you, I wasn't sure whether or not we were going to be able to get through it because it was just so much stress. Thank God we did. But, yeah, that's a very good question. It was a hard dark two years, two to three years there.

Daniel Levine: You were finally pointed to a pediatric endocrinologist. How did you come to see an endocrinologist? And when was that and this progress?

Shirley Dicks: When Elle turned three years old, she still only weighed 20 pounds. So, she still hadn't gained much from the time we started seeing doctors until she was three, and then that summer she had only gained a few more ounces. At that point at our follow-up visit in August, the GI doctor at the time said, "Hey, look, I feel like I'm failing you. She's still not growing. We're not getting anywhere. You guys have done all the leg work, any progress she has made it's because of you guys." He said, "I think you either need to go see a genetics doctor again, or another endocrinologist." So, we decided to go see an endocrinologist. We actually had a recommendation made by a family member of an endocrinologist that specialized in growth issues. We made it a point that we wanted to see

that particular endocrinologist, the downfall to it was he was actually out on medical leave for like two or three months. So, it was going to take some time to get in to see him. But Jon and I talked and we said, if he specializes in growth issues, then he's going to be worth the wait if someone can give us some answers. So we did, we got to see him in October. I think it was October 10th when we got to finally have our appointment with him.

Daniel Levine: And Jon, you seemed to casually mention that she seemed to have an aversion to sunlight. Is that right?

Jon Dicks: Yeah. Yeah, absolutely. That's actually funny that you said that. When we saw the endocrinologist, I'll be honest, when we walked in the room, we knew that something was different here, namely, because we weren't the only ones that were really, really, really worried. I remember it so clearly, he took his little wire rimmed glasses off and he said, "Jonathan, Shirley, I'm really, really concerned about Eleanor. I'm very concerned about Eleanor." He just didn't mince any words there and he went through his reasoning on what was going on and we kind of talked and we talked about the feeding issues and he looked at her and he noticed that her cheeks were a little bit swollen, it looked like she was holding a little fluid to him.

Of course, to us like the normalcy bias there just kind of takes all that away because you see it every single day and you don't think anything's really that wrong, but this fresh set of eyes, I mean, he was like a hawk. He was right on it. And everything about his energy just told me I needed to just listen to this guy and see what he has to say. So, we kind of talked through this and I remember it very clearly, the nurses there, where they were going

to do a blood draw. We were getting Elle prepped for that and we did a good job of letting her understand what that looks like. And she had been through a lot anyway with the pricks and all this stuff. The nurses were like, what do you need? You know, you need like a cup to maybe, or a hat, right. You can go to the bathroom and pee in this hat. We need a little urine. And the joke was that she had been potty trained for like a year and a half before that because of how frequently she had to go to the bathroom. And so, there's this little girl that looks even smaller than she actually is because the kidney disease has affected her at that point in time. It just blew everybody away. And she just looked up and she was like, "No I can be on the toilet. I'll pee in the cup." So, with Shirley's help, they left the room and that's when the endocrinologist and I were in there by ourselves.

And we were talking about it. And to your question, there had been this nagging thing that had always been an issue with Eleanor. And it was something that just seemed so odd to me. And so, I said to him, "You know, doc, I don't know if this has anything, I don't know if this is everything, but I feel like Eleanor has this weird aversion to sunlight. And I mean, really, really weird aversion of the sunlight." And I'll never forget it. The color in his face dropped out. He looked right at me and he said, "you said this, not me. You said that." Right. And I was like, yeah, I said that. And I at first was kind of taken aback because I thought I said something wrong. I thought maybe there was something where I overstepped my balance. I didn't know. And right as I was kind of looking at them a little odd, like what's going on here surely, and I walk back in. And he explained to me what one of the telltale

symptoms of cystinosis is, and it is photophobia or an aversion to light, sunlight, any sort of light. As Elle was growing up, she always preferred a rainy day with puddles over sunlight. Sometimes, it was like, "Mom, can you turn the lights off in the house or wearing sunglasses inside?" And so, for him that was really kind of his confirmatory diagnosis. I think when we left the office that day, we had a very, very clear understanding of what we thought was going on. That's for sure.

Daniel Levine: At what point did you get a confirmed diagnosis and what was it like? Do you have a name to put to this condition?

Jon Dicks: The way the hospital system works is when you give that blood and something is coming down the right, so we understood that after giving the blood work, we were going to get a call from a very, very, very nervous resident that had never seen anything like this before, and like clockwork, later that night at about eight o'clock, we got a call that we have to get in there right now. This time around, rushing into the hospital wasn't as panicked and wasn't as dire straits, because we felt like there was actually a real path that we're following now. You kind of feel like you're on this boat in very, very choppy waters and, I'll be honest, it's a storm, right? There's a storm going on and you have no compass and you're closer to the shore and there's no lighthouse. So, what that diagnosis did for us, and we got the confirmed diagnosis later on, when we came into the hospital that night and the next couple of days that we were there, it meant everything. It's like all of a sudden you have direction and you forget just how necessary it is to have a point to get to and then just a way to try to get there—a compass to try to

navigate these crazy waters that we're in. For the first time it was like, okay, we're actually going to start trying to do some things to actually help Elle, to actually help the symptoms that she has, to help her with whatever's going on right now.

Later on, we learned that everything that she was going through was because at that point in time her kidneys were starting to fail. and she had what they would call in the hospital metabolic acidosis, but it's a symptom of cystinosis and essentially it makes these kids feel like they're breathing under water. And I can remember so many times have little moments in my mind, kind of thinking back to when I was bathing her and she would suck the water out of the washcloth—these heartbreaking moments of where she's trying, without words, trying to give us like, "hey, this is what's going on. Help me, help me," you know? And finally having that diagnosis it reminds you. Just getting the diagnosis started the whole thing. And it's like the actual hard work actually started then because I think there's a misnomer that once you get a diagnosis, everything's better. Well, you still have to get her to that point where she's stable now. So just because we had the diagnosis didn't mean that everything was fixed, but it meant everything to us. I mean, it was scary. You have that moment where you go home and you get on PubMed. At least I got on PubMed. I was a student at that point in time, so for me, that was very high level, but everybody else, what did they hop on—WebMD? You get on Google and you see these horrific stories and you, and you see the worst-case scenarios and you feel like, oh my God, what am I going to do? But after that panic kind of subsides, I think we were kind

of galvanized in fire for the last three years before this, because we had our moment. But I sat down with Shirley and we both looked at each other in the eyes. We said that whatever happens, we are going to advocate for this little girl. We were going to do everything we can to give her the best care moving forward. And we're not going to stop until we get this thing cured and that's it. And so that was our plan. How to get there was all about compartmentalization. So, our big thing was, what's the next best right step?

Daniel Levine: Well, for listeners not familiar with cystinosis, what is it, how does it manifest itself and progress?

Jon Dicks: I could get really medical on the jargon, but I don't think that that's what this is about. It's a rare, a very rare, you could probably classify it as an ultra-rare genetic disease, and there's only 2000, children, actually 2000 people worldwide that have this. It's a lysosomal storage disease. The way it was described to me, and the way I remembered in nursing school, the lysosome is like the trash compactor of the cell. And in this patient population, they have a genetic mutation on this little CTMS gene, all it means is that there's this tiny little mutation. What that means is that the person taking out the trash forgets to take out this one piece of trash, they take out everything else, but they don't take out this one piece of trash. And that piece of trash that's left over, it's called cystine, and they form crystals in the body. Crystals are great in rings, they're awesome outside of the body, but inside of the body they play havoc because they're sharp and they don't look like a cell and they cut through everything and they end up just creating a ton of havoc. So, in this patient population, the cystine crystals accumulate everywhere there

is mitochondria or a cell, and there's not one part of the body that isn't affected: the kidneys, the eyes, the muscles; you get myopathies as well. I think the best way to describe it, the way that I think about it, is it's like this octopus, right? I came up with the octopus because I was trying to think of something that has more arms than me, more arms than me and my wife combined. And that's what it's about. It's like the minute you think you have a little piece of this disease fixed, the other arm part starts creeping up and starts talking about, oh, well, yeah, you've got the kidney issues fixed, but, what are you going to do about the eyesight? And that's the other big thing here. And, like I said before that the kidneys are the windows to the soul, the true rule-out scan to say this is the cystinosis is something called a Slit Lamp Test. The BI docs come in and they shine a very specialized type of light and what you'll see in these kids is that the back of their eyes have a certain shimmer to them. And that's the cystine crystals that deposit in the cornea. Before there was any treatment for this, and I have to be honest, the outlook to this was very bleak and a lot of kids that were born with this just 50, 60 years ago didn't make it past the age of 20. I can remember reading stories like that. Unfortunately we did have a run in with an overaggressive resident that basically made a comment that was just like, we'll be lucky, we'll be very surprised if she makes it to XXX this amount of age. And I was just blown away by it. You know, it's very, very hard to hear someone talk about your daughter as a case. That was one of the things that you have to do a lot in the rare disease community is you have to remind people that this isn't a case number. This isn't an MRN number. This is actually a real person

who has dreams and wishes and hopes and fears. And that was one of the things that we really worked on, worked hard on: making sure that people understand the person behind this disease is really the important part of this, not the disease.

It was a lot to take in and learning about cystinosis and moving forward was really kind of a life's passion. It was something that I put to myself to try to understand as much as I could. So, I utilized everything I could, as far as my academic sources, my academic resources, the other physicians that are around me. I asked a lot of questions and just really surrounded myself with as much material and evidence as I could to try to make the next best step.

Daniel Levine: You found your way to a cystinosis center of excellence in California eventually. What has that meant for Elle's care and her health today?

Jon Dicks: What really was the driving factor in that was the fact that we had for the first time at our institution where we were at, we had a team that was on board that was the correct team that was on board, right. It's not to say that the GI docs or the tummy docs weren't important there; it's just that they shouldn't be steering the ship. Once we had the nephrologist on board and the endocrinologist, we really had the two captains at the helm that were really steering this thing to make things better.

Shirley Dicks: We found the doctor in California after we found a couple of the cystinosis support groups, and someone recommended to go out to see him, to kind of get Elle situated, because even after her diagnosis, we were still struggling with her electrolyte balance, getting that figured out, and

she was still throwing up a ton. And our team of nephrologists was new to this diagnosis so they were still trying to figure things out and how to help Eleanor as well. They were actually on board with us going to see the nephrologists out in California and actually had connected with them through email and things, and still continue to this day when we have questions that we can't quite figure out. So, when we went out to California, it was actually myself, Elle, and at that time, Finley, because I was pregnant when we got Elle's diagnosis—he was only three months old. We went out there to speak with him and he went over I think he said Elle had a thousand pages of medical reports. He went through every single bit of it and he helped lead us on next steps to help get Eleanor through these next few months to try and get everything balanced out. He recommended some things as far as feeding and medication administration and other medications to try, so we brought that back to the doctor and he connected with our doctor as well, and we got all those things put in place. And once we got those things put in place, then we finally started seeing some stability in Elle.

Daniel Levine: What has that meant for her health?

Shirley Dicks: I'm not going to lie, these first few years of diagnosis have been extremely hard, especially the first year, because it is literally a balancing act. She was going in for blood work pretty much like once a week. And then we would eventually get to once every two weeks, once a month, but it was a lot of blood work because you have to find the right dose of medication for her to balance her electrolytes out and to get persisting levels below where they need to be. The medication makes

her very sick so it has to be done in small increments until her body can handle it. It takes a lot of time. I mean, it took months, months. It could have been even up to a year that Elle was able to handle the amount of medicine that she needed without feeling so sick. And even to this day, anytime that we have to increase medication, which you do every time they grow or gain weight, she still struggles, so we always have to do it in very small increments over an extended number of days before we can get her up to her new dose. She's now six years old and she is actually very stable. She is in stage 3 kidney failure. We found that out pretty much right after diagnosis when we were admitted to the hospital, so that is something we have to keep a close eye on.

Daniel Levine: Yeah, forgive me. What is stage three kidney failure. So,

Jon Dicks: There's certain benchmarks that you would see as far as the filtration rate of what the kidneys put out and they give you a rating scale on how to go and where at the time of diagnosis. Unfortunately, because of the two and a half years of misdiagnosis of her, of her being in metabolic acidosis like that, and being in such a harsh environment without the cystine crystals reducing therapy on board, it got her to the appointment. And right now it usually means stage 3 is roughly right around a little bit less than 50 percent kidney function in both kidneys, the right and left are almost the same. One's a little bit better than the other but you can't deny the fact that it is a very aggressive stage of kidney failure for her age, right? At six years old being in stage 3, the writing on the wall there is going to mean that Elle is going to need to have a kidney transplant much earlier than she would have had it had she been diagnosed as

most kids are right around the first year of life—usually about nine months to about 12 months is the kind of the standard, the average of what it looks like. Elle was past three years when she got her diagnosis, so the direct causality there and what you see there is the lowered kidney functioning.

Daniel Levine: You mentioned you had another child since. What's day-to-day life now caring for Elle, doing your job, managing finances? Have things settled into a routine? Is it something that you get more help with? Or how do you manage all that?

Jon Dicks: Well, we have gotten very adept at finding our tribe, finding the people that we can really, really count on. It stands to reason that when bad things happen, you kind of find out who your real people are when the ship's going down and some people are the ones that we knew were going to be there. And some people, we didn't think that we would get this level of help from, and commitment from, the day-to-day life, everything kind of goes on, the big thing. And for anybody out there listening, Finley, our son, does not have cystinosis and is not a carrier for cystinosis either. When Elle was diagnosed, Shirley was six months pregnant. And you can kind of imagine what that looked like for the last three months of that pregnancy. But by the grace of God, we got really, really good news there and Finley is a wonderful little boy who just adores his big sister. It's interesting, we say big sister, but he's probably a little bit bigger than her now and he's two. But the day-to-day interaction there is that he sees everything that we do with Elle, he understands what else is happening with her. As far as the day-to-day stuff, Shirley's still working part-time, I myself decided that I

was going to get out of the ER and have since finished all of my work to become a nurse practitioner. From that standpoint, I'll sit for the family nurse practitioner exam probably in December of this year. Right now, I own and run a personal training, fitness coaching business. The time spent there is a little bit different now. I just really realized that I couldn't do that level of care for people with what was going on with Eleanor and I had to be very honest with myself that I was really running myself into a big pit of depression, and we all kind of pulled ourselves out doing what we knew we needed to do, and that's just surround ourselves with really, really good people. We know that the bills are always going to be there. Every month, somehow, we figured out a way to make it happen, with the faith and the knowledge that if we keep on doing the next best thing, that good things are going to come. Our driving goal here is always just to try to make sure that we reduce any undue stress on Eleanor as much as we can.

Daniel Levine: I imagine the whole diagnostic odyssey for you was very isolating. What's it like now to have become a member of a community around this condition?

Jon Dicks: It means everything. And it's not even a new thing. When before you know it, there's a community of people out there that understand the same thing. Like when you find someone that understands what it means to get popped in the face with a G-tube with the medication that you're pushing, like, you really don't know. Right? I mean, you can talk about this kind of anecdotally, and you'd be like, oh yeah, the G-tube popped on me. But then when you're dealing with a family that actually has a son or daughter with a G-tube, somebody that is presenting the same

way, someone that understands kind of what that cyclical vomiting looks like, and what fractured sleep is like what.

Shirley Dicks: Medication.

Jon Dicks: Right, right. You know, the weird alarms that go off six times a day, and people are like, what's that for? It's just like you found your home and you realize that you can really utilize that home, that second home as a source for an abundance of strength. You see other people that have gotten through this, you meet other people that are in their 20s, 30s, 40s, 50s, and you're filled with this hope because as a father one of the things that always gets me really choked up is the thought of possibly not being able to walk my daughter down the aisle, right? This crazy thought that somehow, I would outlive my daughter and then you see that there is life after cystinosis. Obviously, it's a through line and it drives all the decisions that you make, but there is a way through this. And I remember when I finally got to talk to some parents that that was the big message then said, "you know what you're embarking on is some of the hardest stuff you're ever going to be going through the diagnosis. This early part is going to be hellacious, but if you can get through that and if you can get through it together, it's worth it and it gets much better." And while it's light years away from the normal that you had before, what we realize is that we're dealing with a new normal now. And so that's the way we approach it. Now, our life is not like most, but it's wildly fulfilling and wildly rich. And we laugh a lot. We cry at times too, but we do it together.

Daniel Levine: And has the experience put you into the world of advocacy at all?

Jon Dicks: Yeah, actually it has. I, I remember when Elle was diagnosed, I looked at Shirley and I said, you know, I'm going to learn everything I can about this. I'm going to do whatever I can to be the change that I want to see. And I really did want to see a lot of change at that time, because I was dealing with a daughter at that point in time, what I felt like was going through undue stress because the teams were very myopic in their approach and they didn't connect those dots. So, what I wanted to do is I wanted to figure out how do I speak for Eleanor and speak for all these other kiddos around me that are undoubtedly going to be coming behind? How do I help them? So, initially what we did is I went to my first cystinosis research network conference and that was in Philadelphia. We went to Philadelphia and our minds were blown. Shirley would agree. It was incredible.

Shirley Dicks: It was the best thing we could have done at that time for our family. We were only, I think, six months post diagnosis. And, luckily this conference was being held that summer and it was definitely the best thing we could've done for our family to be there with a group of families who have been through or going through the same thing. They just shared so much knowledge, so much love, and we made so many connections. Jon made a ton of connections. He really was able to reach out and meet a lot of people there. It was absolutely amazing.

Jon Dicks: It was from that conference that I found out that there was the ability to go down to Washington DC for Rare Disease Day. I had no idea that this was even a thing. I was able to get down there as the representative for my district in Ohio where I'm from. I was there and I came with a plan and I was able

to sit down with my congressmen and women and senator as well. And it was just like the world kind of opened up from there. I realized that the advocacy that I could do is much, much, much more than what I thought. Out of that advocacy there, and then the incredible experience that I had at the conference, I was actually asked to come on and voted onto, the executive committee of the CRN. And I've been on as the VP of development for the last year. So, I took that advocacy and I really, really ran with it. It's an incredible position. It really allowed me to speak to other families, which was something that I really, really wanted to do. I felt like, what I really needed in those dark times with somebody who understood what I was going through and was able to kind of help me work through it and work through some of the things and some of the nuanced stuff that kind of comes out of a new diagnosis. Right? So, it was a wonderful opportunity for me to be able to meet with industry professionals, to really get my head wrapped around the research. You want to do things that directly affect the community so being in charge of research advocacy, of working on securing financial assets for these incredible doctors who are doing incredible work around the country and around the world, was amazing. And I really fell into the role. About a month ago, I was asked to come on board as the president of the CRN. So, right now I'm the acting VP of development, and then I'll also be taking over as the president of the CRN. So, I definitely am advocating that's for sure.

Daniel Levine: How's Elle doing today?

Shirley Dicks: She is doing very well. We actually got bumped to six months nephrology appointments. That's how

well she's been. She's been stable and everything's looking well. We still do go for 3-month blood work just to make sure we don't have to make any tweaks or if we do have to make any tweaks in her medication. The only big change is we've had to add a urologist on board and now she has to have a kidney scan every three months, just to keep track of her kidneys and make sure they're filtering out okay and not slowing down even more. We're keeping a closer eye on that. But as long as everything stays okay with that, she will just continue to have kidney scans every three months. And her nephrology appointments will stay every six months along with our ophthalmology appointments, our eye appointments. So far, she's been healthy and we started homeschooling. She's kind of enjoying that. I guess it's a new thing for her. We're still getting in the groove of that, but she has tons of energy to be outside, still in the rain. She loves her bugs. She loves playing with cousins and friends. So, we're very, very happy and very pleased with how well she is doing.

Daniel Levine: Jon and Shirley Dicks, parents of Elle with childhood cystinosis. Jon, Shirley, thanks so much for your time today.

Shirley Dicks: Thank you so much.

This transcript has been edited for clarity and readability. Listen to the full RAREcast interview here:

<https://bit.ly/3Am5mLe>.

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 afán 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 tiroidea

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.



3141 Caniff Street Hamtramck, MI 48212

info@BrombergTranslations.com

Phone: 313-871-0080

Fax: 888-225-1912

www.BrombergTranslations.com

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



3141 Caniff Street Hamtramck, MI 48212

info@BrombergTranslations.com

Phone: 313-871-0080

Fax: 888-225-1912

www.BrombergTranslations.com



Rare Dedication

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.



Research Committee Update

By Christy Greeley, Vice President of Research, Executive Director



From left to right; Jack, Christy and Alex Greeley.

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 and does so currently. 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. We look forward to issuing another call for research proposals in 2022.

Current CRN Grant Commitments

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 Levtchenko

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

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

function, extra renal complications and a WBC cystine-based long-term compliance score.

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis

Katharina Hohenfellner, MD

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

The concept presented here, follows the established approach for the rehabilitation of children “Auf die Beine”, which was developed in Cologne for children and adolescents with limited mobility, e.g. caused by cerebral palsy or patients with osteogenesis imperfecta (<https://unireha.uk-koeln.de/kinder-jugendreha/behandlungskonzept-auf-die-beine/>). The training design with home exercise and short training sessions considers the fact that the target patient group is already under great strain due to the severity of the disease (medication, special diet, possibly dialysis). The project follows the official recommendations for the use of a vibration plate (4,5). Patients train with Galileo vibration plates according to a fixed training schedule which provides for 10 short training sessions per week (maximum 2 per day). Within one training session, four exercises will be performed. The control group will perform the same exercises without vibration plates, but with dumbbells. Patients will initially undergo an intensive training course and will receive regular supervision during the three-month home training phase. The study is designed as a randomized controlled trial. Due to the very rare underlying disease a matched pair design was chosen to achieve comparability between the two groups. The patients will be matched based on their age, sex, and major previous surgeries. A baseline and two follow-up clinical

assessment, one after the three-month home training phase and one after the follow-up phase, will take place. The primary endpoint is the change in muscle strength (in %) from the baseline examination to the measurement after the training phase. In the control group (CG), a mean shift of 0% is expected, since no vibration training is carried out.

Potential Impact for patients with Cystinosis:

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

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 is 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



Arndt/Greeley family time.

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 aminothioliol cysteamine, which is highly effective in reducing the intracellular levels of cystine. However, this drug is ineffective in the treatment of male infertility in these patients.

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

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

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

allow Dr. Sur to approach cystinotic patients for access to patient biosamples.

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

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

The Specific Aims of this project are the following:

- Aim 1: Generate human immortalized CTNS^{-/-} epididymal and testis cell lines by CRISPR/Cas9 and confirm the phenotype to further downstream study of male fertility associated with cystinosis
- Aim 2: Map the molecular perturbations in both cell lines with deletion of CTNS and in tissue samples from male cystinotic patients, by using state of the art genomics that the Sarwal Lab has legacy expertise-in. This will define the clinical utility of the resource generated in Aim 1.

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

Cystinosis Community Advisory Board

I continue to participate as the U.S. representative in the Cystinosis EuroCAB programme, a project of EURORDIS, the European Rare Disorder Organization. The Community Advisory Board's (CAB's) objective is to improve patient access to novel therapies and treatments.

This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research. We look forward to 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. Look for an expanded Dialysis and Transplant section coming soon which will include a broad range of information and resources for those facing these challenges.

Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from around the world. Also be sure to check out the many cystinosis related articles and publications available in our Publications and Guides library at <https://www.cystinosis.org/support-resources/publications-guides/>

CRN Financial Review – Treasurer’s Report

By Tim Wyman, Treasurer

We are in the final stages of our annual audit (2020) conducted by an independent accounting firm in accordance with generally accepted auditing standards. As CRN has done in the past, Form 990 will be posted upon filing at <https://cystinosis.org/about-us/financials/>. Some of the financial highlights include:

- Total Revenue from sources such as grants, contributions, and fundraising events were \$527,497.

- Expenses incurred for the year were \$200,789. This was lower than 2019 as 2020 was a “non-conference year”.
- Net assets grew by \$326,708.
- Grant commitments for research projects totaled over \$376,000 and in early 2021 CRN awarded 3 additional grants totaling approximately \$436,000 to be distributed in installments and based on satisfactory progress reports.

CRN is well positioned as a not for profit organization and resources are dedicated to supporting and advocating research, providing family assistance, and educating the public and medical communities about cystinosis.

Lastly, a special thank you to volunteer Jenni Sexstone for her past contributions as CRN Treasurer. As a volunteer organization, the efforts of community members like Jenni are invaluable and we appreciate her contributions to the CRN community!

“ Thank you for helping people! ”

– AYSHA ALLIE HAGENE

“ Thank you so very much. I never expected this to happen today. Truly grateful and thankful for this opportunity. I appreciate all you have done. ”

– LINDA AND LUCAS STONE

“ Awesome! Thank you so much Jonathan! ”

– KATIE KUNDE

“ On a long stay, our computer lets us stay in touch with family, and do video chats! We appreciate you for giving us a chance to do that! ”

– NICOLE HYATT



pcsforpeople

Now available for individuals and families living with cystinosis. PCs for People will provide a personal computer and internet connection. Contact Jon to take advantage of this program: jdicks@cystinosis.org.

“ We received the laptop today its AMAZING THANK YOU SO MUCH MERRY CHRISTMAS AND HAPPY NEW YEAR! ”

– DEMERE OAKLEY

“ I just wanted to follow up with you and thank you for helping me. Jackson has the computer and loves it! ”

– JEAN BLUM

“ I can just say that I will do what I can to aid in funds being allocated to your nonprofit as I find it genuinely just beyond kind. I have never been offered such kindness to try to offset my suffering and aid me in driving my life in a positive direction. ”

– ROBIN POWERS

“ Wow!!! Thank you and congrats on making such a wonderful impact on the community, kudos to you!!! ”

– JENNIFER BARTKOWSKI

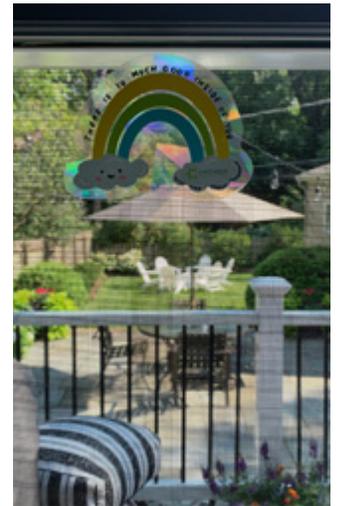
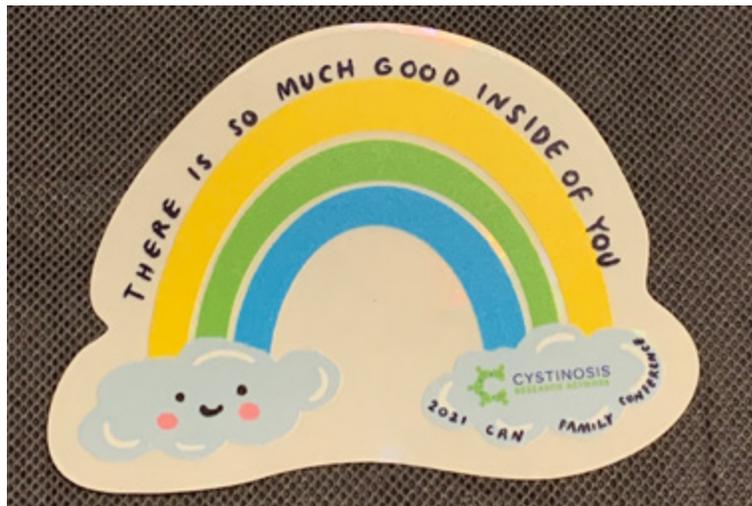
Join the Cystinosis Research Network

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.

What is the purpose of this program?

NORD's Cystinosis Patient Assistance Programs offer eligible individuals diagnosed with Cystinosis financial support when faced with limited resources to pay for:

- out-of-pocket healthcare costs, and/or
- diagnostic testing costs, and/or
- unexpected emergency expenses

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

These programs are designed to help patients who:

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

What kinds of assistance can I request from NORD?

NORD's program can assist eligible individuals/families with expenses in a number of categories:

- The Cystinosis Copay Program assists eligible individuals who have health insurance with funding to cover health insurance deductibles, copayments & coinsurance costs associated with the care of Cystinosis.
 - Some examples of these expenses may be:
 - medical expenses paid toward health insurance deductible
 - copayment for a medical office visit
 - out-of-pocket cost for an Emergency Room visit
 - a coinsurance payment for a consult with a Cystinosis specialist
 - This program does not assist with copayments for medications
- The Cystinosis Medical Assist Program assists eligible individuals who are uninsured, or for whom coverage has been denied with out-of-pocket costs for medical expenses such as medical visits, laboratory & diagnostic testing, wound care products and other specific medical expenses. Additionally, mileage for travel to and from a Cystinosis related medical appointment may be reimbursed in this program. Medication costs are not covered.
- The Cystinosis Emergency Relief Program assists eligible individuals with the cost of unexpected or emergency non-medical expenses that cannot be afforded without short-term assistance.
 - Some examples of these expenses may be the cost of repair for a car or major appliances, unexpected utility costs or cell phone bill that cannot be afforded due to lost wages from time off related to care of ill family.
 - Other types of emergency requests will be considered on an individual basis.

FAQ

What is NORD?

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

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

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

Is there a fee for applying for assistance?

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

NORD does not recommend or endorse any particular medical treatment but encourages patients to seek the advice of their clinicians. Donations to NORD for this and other programs may be made by contacting NORD at rarediseases.org.

What is the application process?

Awards are granted on a first come, first served basis. Patients may be referred to the program by their health care provider, their case managers, or they may self-refer. The RareCare[®] Patient Services Representative will guide the applicant through the application process, verify eligibility for inclusion in the Cystinosis Program(s), determine financial eligibility using our Electronic Income Verification System (EIV) and award assistance.

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

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

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

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

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

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

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

Phone: 855-201-5087

Fax: 203-486-8033

e-mail: cystinosis_assist@rarediseases.org

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

US MAIL to: NORD

Attention: Cystinosis Program

55 Kenosia Avenue, Danbury, CT 06810

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

Copay and Medical Assist awards are issued for a calendar year.

Emergency Relief awards are offered as payment support for a one-time capped emergency need.

How does the payment or reimbursement process work?

Copay and Medical Assist awards may be prepaid by NORD with appropriate documentation or reimbursed to the patient in accordance with appropriate receipts and documentation.

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

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

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

Are there expenses which cannot be covered by NORD'S Emergency Assistance Program?

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

- Federal, state, or local tax payments, including property taxes, child support payments, legal fines and/or fees
- Luxury goods and services or vacation costs are not eligible for consideration

NORD does not recommend or endorse any particular medical treatment but encourages patients to seek the advice of their clinicians. Donations to NORD for this and other programs may be made by contacting NORD at rarediseases.org.

CRN Year End Giving Campaign

As 2021 winds down, please consider donating to our year end giving campaign. The CRN is the only cystinosis advocacy group providing research funding, individual and family support, and education and awareness. Your donation can help accelerate the discovery of a cure, the development of improved treatments and enhance the quality of life for people living with cystinosis. Visit cystinosis.org/howtohelp-donate to make a tax deductible contribution today.



MEET RAYAN

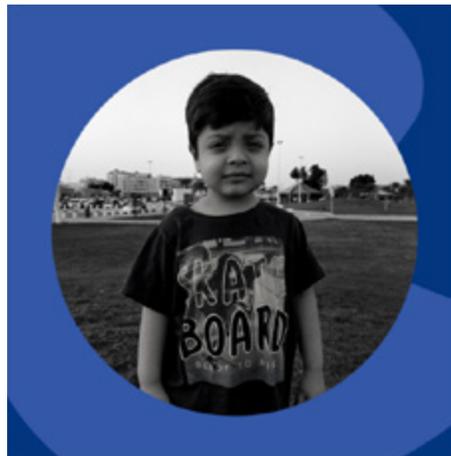
It is one rare disease which brings a lot of discomfort to us but we as a family, for our kids will fight against the disease and our spirit and trust will take us on the other side where we will see our kids thriving. Have faith and stay strong.

*"You are one unique blessing of Almighty Allah. You are strong and everyday that we spend with you is worthy and exciting. The future is yours and as a parent, we will never let you fall.
Sincerely, Mama and Baba."*



MEET ANNICA

When we got the diagnosis my world turned around and I thought we would never make it work. But with knowledge, love from friends and family and support from the cystinosis family we have made it so far. Now we hope for a cure and a better future for all living with this disease.



MEET JONATHAN

Some say why me? I say why not me. Cystinosis is a very dangerous and harmful disease. Although everyone deals with something in their life. Some have it better and many have it worse. Count our blessings

"Rare diseases are tough please don't judge others"



MEET ISAAC

We are getting closer and closer to a cure!

*"Isaac keep doing you! Remember your capable of anything
Dad and Mom"*



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

PRESRT STD
US POSTAGE
PAID
KANKAKEE IL
PERMIT 38

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



CYSTINOSIS
RESEARCH NETWORK

Volume 14, Issue 2
Fall/Winter 2021

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

Phone 847-735-0471
Email info@cystinosis.org

Editor Clair Johnstone

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

CRN VISION

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

CRN MISSION

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