After a year of planning and endless preparations by so many, the time finally arrived for the 2023 CRN Family Conference held at the Vanderbilt Marriott in Nashville, Tennessee. This marked a 20-year milestone for CRN, and the 11th biennial conference CRN has sponsored. Tee-shirts showcasing 10 of the conferences were on display near the registration area along with a map inviting attendees to “pinpoint” their home location both nationally and internationally. It truly was a global event with attendees arriving from Ireland, Mexico, Netherlands, Norway, England, Belgium, Egypt, France, Germany, Italy, Scotland, Turkey, Canada, and the United States. In total 33 states were represented.

Continued on Page 5
Dear Cystinosis Community,

I hope this letter finds you well and full of renewed hope after our incredible time together at the 2023 Cystinosis Research Network Family Conference in Nashville, Tennessee. As I sit down to reflect on the moments we shared during this past July, I am overwhelmed by a profound sense of gratitude for each and every one of you who contributed to making this event an exceptional success.

Our first in-person family conference since the challenges of the past two years was a heartfelt reunion that far exceeded our expectations. The opportunity to connect face-to-face, to embrace one another with open arms, and to witness the strength of our community was a testament to the resilience that defines us. Stepping into the conference halls and seeing familiar faces, as well as new ones, filled me with immense joy.

In the spirit of building upon our shared experiences and fostering growth, the conference agenda was carefully curated to address the diverse needs of our community. The conference covered a range of topics, from the latest advancements in cystinosis research and treatment to practical workshops that empowered families with the tools they need to thrive. We celebrated breakthroughs, explored innovative therapies, and delved into the future of cystinosis care.

The moments we shared were filled with inspiration and hope. The litany of incredible speakers left us with a profound sense of possibility, reminding us that through unity, we are capable of achieving greatness.

Our time together not only enriched our understanding of cystinosis but also deepened the connections that make our network so strong.

One of the highlights that I cannot help but mention is the incredible sight of both familiar faces and new ones at the conference. It was heartwarming to see our established families and patients reuniting with old friends, sharing their journeys, and offering support. Equally heartening was the sight of so many new faces, individuals who are now woven into the fabric of our community. The growth and inclusivity we witnessed speak volumes about the power of our shared mission and the welcoming nature of the Cystinosis Research Network.

I want to express my deepest gratitude to everyone who made this conference possible - our dedicated team, generous sponsors, passionate speakers, and, most importantly,
The President’s Letter, continued

each member of our community who attended. Your presence and active participation made the conference a true celebration of progress, strength, and unity.

As we move forward from this remarkable event, let us carry the spirit of the conference with us. Let us harness the knowledge gained, the connections made, and the inspiration felt, to fuel our ongoing efforts in driving cystinosis research, support, and advocacy. The road ahead may have challenges, but with our united force, we are equipped to overcome them.

Thank you for being an integral part of the Cystinosis Research Network family. Together, we are making a meaningful impact and fostering a brighter future for all those affected by cystinosis.

With heartfelt appreciation and optimism,

Jonathan Dicks

NIH Rare Disease Day & RDLA Rare Disease Week on Capitol Hill

By Jonathan Dicks, President & Vice President of Development

Fresh off of a wildly engaging opportunity for not only rare policy advocacy but the simple act of in-person fellowship, I finally have a moment to digest it all. This was such an impactful trip for me in so many ways. It seems like a lifetime ago our family was fresh off the high of “finding our team” in Philadelphia at our first (and only) in-person CRN Family Conference in 2019. I was motivated and ready to serve. I was invited and agreed to accompany a select group of cystinosis patients/advocates to Washington, D.C. to attend my first Rare Disease Week in 2020.

We had just found others that understood what living and caring for someone with this rare disease felt like. It was a tiny group of ardent warriors whose spirit and resolve was something I had never seen before. It wasn’t until I attended my first RDW that I got a clear understanding of just how many rare warriors truly existed. 30 million Americans are living with one or more rare diseases, a number now estimated at over 10,000+ distinct rare conditions, of which, only 5% with an approved
NIH Rare Disease Day & RDLA Rare Disease Week on Capitol Hill, continued

benefit is far-reaching for those in and outside of the rare disease space; citing the landmark Burden Study that estimates the 2019 impact of rare diseases in the U.S. at nearly $1 trillion!

We lobbied our respective Senate and House members to join the Congressional sign-on letter to the FDA requesting the formation of an internal FDA task force to review agency-wide rare disease activities and requested co-sponsorship of the BENEFIT (Better Empowerment Now to Enhance Framework and Improve Treatments) Act requesting the FDA to include patient experience or patient-focused drug development data as part of its risk-benefit framework. This tool for evaluating risk-benefit does not currently have data from the patient perspective that could be critical to informing the evaluation and, ultimately, decision on whether or not to approve a product.

We asked our elected officials to consider joining the Rare Disease Congressional Caucus, a bipartisan group working to raise awareness of rare diseases by helping bring public and Congressional awareness to the unique needs of the rare disease community (including patients, physicians, scientists, and industry), and creating opportunities to address barriers to the development of and access to life-altering treatments.

We began the week in Bethesda, MD at the NIH campus for a day full of presentations that highlighted NIH-supported rare disease research and the development of diagnostics and treatments. We had mutually beneficial dialogue among the rare diseases community while exchanging the latest rare disease information with key stakeholders, advancing research and therapeutic efforts. The day was capped off with an incredible array of rare patient personal accounts and a lovely reception at the Ronald Reagan International Building followed by a rare disease feature film screening.

We then got to the business of rare patient advocacy, partaking in breakout sessions discussing the pertinent federal issues directly affecting the rare disease community. We heard discourse on policy issues stressing the extreme importance of the continued investment in rare disease research, noting how the

voice, and when magnified as a collective we can and will change policy and save lives. I was honored to represent the Cystinosis Research Network and the great state of Ohio during my time in Washington D.C. and know how impactful we can be when we speak with one voice.
Near the registration area attendees could meet and greet John and Susie McCalla as they handed out Cystinosis themed comic books created by their late son, Kevin. They could also learn more about the “Live Like Laura Fun Fund” scholarship started by Frankie McGinnis in memory of her daughter Laura McGinnis. ALAB manned a table too, answering questions about the adult cystinosis community and handing out ‘goodies’.

There was a palpable sense of excitement, anticipation and connection as attendees arrived to check-in. It had been four years since the last in-person conference. (Due to Covid concerns the 2021 conference was virtual.) Old friends reconnected with hugs, smiles, and conversations, happy for the opportunity to meet again in-person this year. Researchers, doctors, and industry partners also gathered to share, learn, and be a part of this special experience that can’t be described only felt.

Surrounding the main gathering space in the foyer were many posters targeting current research, quality of life issues, medications, international connections, and opportunities for adults who live with cystinosis to become involved.

On Wednesday, July 12th, the CRN Board of Directors met in an all-day session to discuss the direction and future of the organization. They also elected two new board members for the 2023-2024 term, Heather Rothrock and Terri Schleuder.

A welcome reception for “Speak Up Speak Out” sponsored by Horizon Therapeutics for young adults and teens who live with cystinosis was also held. This two-day groundbreaking, spoken word, self-advocacy workshop focused on empowering participants to use their own words and perform publicly sharing their ‘truth’ through the creative arts.

On Thursday July, 13th, from 9:00 am-4:00 pm a Scientific Symposium was held providing an opportunity for doctors and researchers to share their latest efforts and discoveries. Topics shared included: Pathogenesis and New Interventions, New Pharmacological treatments for cystinosis beyond cystine accumulation, Potential of RNA therapy in genetic diseases,
Early diagnosis and biomarkers, and newborn screening. The symposium also addressed advocacy and quality of life issues such as fertility preservation and new strategies in kidney transplantation.

This year, we had a large number of first-time attendees who were welcomed with an orientation, networking session hosted by Jonathan Dicks, President and VP of Development, and Jen Wyman, VP of Family Support.

The first “official” event of the conference was a welcome reception and complementary dinner hosted by CRN’s Board of Directors. It was an informal opportunity for families to greet each other. This was followed by more formal family introductions, which provided an opportunity for families to introduce themselves, state where they were from and share their connection to cystinosis.

On Friday, July 14th, it began. Jonathan Dicks, President and VP of Development and Christy Greeley, Executive Director and VP of Research welcomed attendees with personal stories, connections, and a description of CRN as an advocacy group.

Titled Cystinosis 101, Bill Gahl, MD, provided a detailed overview of cystinosis. This was an excellent review, especially helpful to those still learning about the disease.

Paul Grimm, MD, followed with an informative discussion of the kidney transplantation process. He described what the stages of chronic kidney disease (CKD) mean, labs that measure kidney function accurately and the pros and cons of living donor vs cadaver transplantation.

The process of designing clinical trials needed to bring new therapeutic treatments to market was discussed by J.J. Zaritsky, MD, including therapies involving genes. Stephanie Cherqui, PhD, discussed updates in the clinical trial of stem cell gene therapy for cystinosis. Of the six patients who have received the treatment five no longer require cysteamine depleting therapy including Jordan Janz, patient one. Jordan, was also available to describe his experience with the stem cell treatment which he received four years ago and his follow-up since. Hearing him describe the good and bad of the experience, and to hear how good he feels now without cysteamine therapy was heartwarming. This clinical study was certainly a highlight and offered hope for the future in cystinosis treatment.

Next, the Speak Up Speak Out: Cystinosis and Me group delivered a heartfelt performance of poetry about their cystinosis journeys. Their “truth” touched us all and brought tears to many eyes.

Two group photos followed. One of all the attendees and a second of those living with cystinosis.

Following lunch, a scavenger hunt through Centennial Park was held for teens and young adults. In spite of the heat, it was fun for all who participated. Simultaneously several breakout panel sessions targeting different phases of the cystinosis journey were held. Attendees could choose to attend the one that most applied to their experience.

Topics included in panel session 1:
Newly Diagnosed Families
Middle childhood and teen years
Adults

For panel session 2 these topics were offered:
Newly Diagnosed and middle childhood, physician Q & A
Transplant and Dialysis
Adults living with cystinosis as an adult (closed session)
Parents of Adults living with cystinosis (closed session)

Both of the closed sessions offered a safe place to voice concerns and be heard by peers.

Closing out Friday’s sessions was another highlight, the Medical Panel, moderated by Dr. Bill Gahl. All physicians in attendance came together for 90 minutes to answer questions from attendees. Seldom are so many experts in one room willing.

It really is incredible how many people are here and how this community comes together so well and has stayed together.

- Dr. Bill Gahl
and able to answer whatever was asked.

Friday evening the Adult Leadership Advisory Board (ALAB) hosted an adult meet-up to informally gather at the hotel. After that several adults who live with cystinosis, continued onto downtown Nashville to enjoy music and fun together.

On Saturday, after opening comments from Marybeth Krummenacker, VP of Education and Awareness our industry partners provided updates.

Horizon Therapeutics- Andrea Atherton, and Tovah Toomasson
Leadiant Biosciences: Cystaran, from research to approved therapy- Lesli King
Recordati, Rare disease update- Anna Vorobeva

Following these updates Denise Dunne, from Cystinosis Network Europe (CNE) spoke.

CNE is an umbrella of groups connecting patient support, advocacy, and research. In addition to hosting a family conference every 2 years in Europe, the CNE coordinates the cystinosis Community Advisory Board (CAB). This is a group of patient representatives who offer expertise to public and private sponsors of clinical research.

The next session, titled Finding our Roots, moderated by Jean Campbell, the former NORD V.P. of Development, and featuring Dr. Jess Thoene, Dr. Bill Gahl and Marybeth Krummenacker was very informative and tugged at the heartstrings of many who have been on this journey for a long time. It certainly was a highlight. These experts discussed the story of cystinosis and their connection to it. It was a story of where we started and how far we’ve come. It was a story that discussed the importance of research, connections, collaborations, government institutions, patient advocacy, clinical study participants and even coincidences. All this melded together over 40 years to form organizations like NORD, the Orphan Drug Act and miraculously 4 FDA approved therapies for cystinosis. We have personally been a part of this journey since 1989 and have seen so much progress outlined by these two remarkable doctors/researchers. (Dr. Thoene requested a moment of silence for the late Dr. Jerry Schneider, another important part of the story.) We are so grateful and humbled and know the next amazing steps toward treating and eventually curing cystinosis would not be possible without what has gone on before. Dr. Thoene mentioned the very first patient to take cysteamine as

The most recent Cystinosis Research Network conference in Nashville was inspiring. As an adult living with cystinosis, seeing the support and enthusiasm in our rare disease community is something I truly cherish. Observing my peers and the younger families thriving with cystinosis is a reality we all have worked diligently for, seeing it all come to fruition is a reminder of how there truly is strength in numbers.

Many of the adult patients are getting older and having a community to laugh and cry with gives us purpose and solidarity.

I am honored to be a part of the cystinosis community and envision a bright future with many more conferences and events for community engagement to come.

- KD, adult living with cystinosis
The next presentation was titled: Meeting the Challenge of Rare Disease in the Family: 25 years of Lessons Learned, by Al Freedman, PhD. Dr. Freedman, a soft spoken psychologist, outlined the common challenges faced by all families with a rare disease. He touched all of us with the connection we all share as part of the larger rare disease community. His “truth” took my breath away as it merged with my own. Dr. Freedman, described his personal experiences as the father of Jack, who passed away from Spinal Muscular Atrophy (SMA) and the lessons he learned. These apply to all rare families.

The Lessons
A rare disease diagnosis is TRAUMA
It is hard to live with UNCERTAINTY
HOPE breeds hope
FAMILY matters

COMMUNITY strengthens and supports
FRIENDS make a huge difference
DOCTORS & NURSES are people too
SCIENTIFIC PROGRESS brings new hope and new challenges
EMOTIONAL help is important too
ANYTHING is POSSIBLE
Rare disease also brings RARE GIFTS
-our families model RESILIENCE
-our families are PIONEERS
-our families attract other people who have COMPASSION
-our families bring people TOGETHER
-our families bring out THE BEST in us

After lunch, Mona Suck, MSc, described her study, QUALIFY-US. This is the development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis.

The day closed with two highly anticipated panel sessions:
Parents of children and adults with cystinosis
Adults living with cystinosis
By using several prepared questions, and opening it up to attendee questions, much information was shared by these “experts” living on this continually evolving journey. Attendees sat on the edge of their seats and absorbed every word from people who know the ‘road’ well. Learning from each other, and supporting each other is one of the main conference goals.

Closing comments were made by Jonathan Dicks and Christy Greeley. The conference concluded with a magnificent dinner/dance. The food was superb, the music lively, and everyone danced the night away. Saying goodbye is hard….can’t wait till next time.

As one might expect, putting on a conference of this magnitude is HUGE, and could not have been accomplished without the help of so many. To the entire CRN Board and Executive Committee, THANK YOU. A very special thank you to Dana Marshall, Clair Johnstone, and Allison Goldberg whose help was invaluable in ALL aspects of the planning and execution of this conference. We would also like to thank Maya Doyle for her continued support of our cystinosis community.

For more photo highlights turn to Page 10.
Our experience as first-time CRN conference attendees was extraordinary in every way. The clinical information presented reiterated things I had learned previously while also introducing concepts I was eager to learn; I could not write notes or take pictures of slides fast enough in some of the sessions. The opportunity to hear from and ask direct questions to the cystinosis specialists was enthralling, knowing that several of them had been involved with cystinosis from its early recognition and played a critical role in the research and development of the treatment options available today.

I was particularly struck by the comments shared and interactions I had with several of the adults living with cystinosis—meeting Jordan Janz as the stem cell trial trailblazer left us feeling starstruck, like we had met a celebrity in our rare disease world. There wasn’t enough time to express to Jordan and the other cystinosis adults (and their parents) what their experience has done/will continue to do for our kids, and the contributions their efforts have made to the evolution of managing this disease.

Getting to spend time with other parents new to the journey like us, some even newer, gave us an indescribable, unprecedented sense of community—though many of us had never met, there is an instant connection and empathy on a level likely deeper than anything else in our lives. On a day-to-day basis, cystinosis can feel overwhelming and it is often difficult to find people in our everyday lives that can relate. However, the conference allowed us in-person time with other families that understand our struggles, establishing lasting relationships that we can lean on for support in the years to come. Not to mention the connections our children are making—Briley is still talking about the friends she met in Nashville; she does not yet understand the full spectrum of what these friendships will mean in the coming years.

As a parent of a young child with a rare disease, feeling like I have connectivity and resources is critical for my child’s physical health, my mental health, and my family’s overall well-being. We left the CRN conference feeling empowered and refreshed with realistic, meaningful access to people and information immediately applicable in our lives. We eagerly look forward to the next CRN event!

- Heather Rothrock, caregiver
2023 Nashville Conference Photo Highlights

Attendees from the U.S. and beyond

Speak up Speak Out participants

Dr. Gahl and Cystinosis 101

Dinner dance fun

Medical Panel Q & A

Daycare fun with princesses
ALAB’s Conference Impressions
By Jana Healy, ALAB Chairperson

The Adult Leadership Advisory Board attended the CRN’s Family conference in Nashville this past July. There was tremendous excitement in the air as we all had the pleasure of meeting in person, some meeting for the first time and others getting to reunite after years of being apart. We were not only happy to be a part of many adult specific sessions, but also seeing many people willing to share their story in order to help and encourage one another was uplifting.

A few of our ALAB board members weigh in with their feelings about attending.

KD mentions: “I could have never imagined in my lifetime we would be discussing the multiple options available to treat future generations to come. On a more personal note, being surrounded by other adults thriving with Cystinosis is a reminder of how far we have come and how we can lead by example to uplift one another.”

Another ALAB board member, Sara, adds: “The best part of the conference was meeting adults in person and learning how to better help our community.”

A third ALAB board member, Steve, explains: “I had multiple parents/families come up and talk to me about how much it meant to them that I and other Cystinosis adults were there at the conference. They explained that we represented hope for the future to them. I can sometimes get lost in my own world and lose sight of my role and importance in the Cystinosis world. It’s good to be reminded of this every now and then.”

Seeing everyone in person, each at different parts of their Cystinosis journey made the conference truly an inspiring and heartfelt event to be a part of.

The ALAB’s mission is to support our community and provide resources through our programs. You can reach out to ALAB through email: alab47213@gmail.com or find us on Facebook.
This is the time of year when CRN’s Board personnel changes. This year it is a BIG change. We send the biggest thank you possible to Christy Greeley, who has been involved with CRN in so many capacities for over 20 years and is stepping down. As a past President, and the current Executive Director and VP of Research, Christy’s leadership has been invaluable in allowing CRN to achieve its Vision and Mission to support, families, fund research, and educate both the public and medical professionals about cystinosis, while always reaching for improved treatments and an enhanced quality of life for those living with cystinosis and their families. Thank you, Christy. You will be so missed.

In addition, Jen Wyman will be stepping off the Executive Committee as the VP of Family Support. She will remain on the CRN Board as a Director. Current Board member, Chelsea Meschke will be stepping into the role of VP of Family Support, congratulations, Chelsea.

Board member, Kristina Sevel, will step into the role of VP of Research as Christy exits this position. Thank you, Kristina.

Clair Johnstone, a long time supporter and friend of the cystinosis community will become our new Executive Director. We are so happy to have you on Board in this new role, congratulations Clair.

Two new CRN Board members were elected at the in person Board meeting held at the Nashville Family Conference. Welcome to Heather Rothrock and Terri Schleuder.

Heather and her family live near Winston-Salem, North Carolina. She became passionate about cystinosis when her four-year-old daughter, Briley, was diagnosed with disease at age 18 months. With the uncertainty and fear at diagnosis, Heather found support and grounding by connecting with families on a cystinosis Facebook page and through CRN. She also met with Dr. Gahl at the NIH, which she highly recommends.

Professionally, Heather has an MBA from East Carolina University and currently works in the Population Health department at Atrium Health-Wake Forest Baptist Hospital focusing on cost and utilization management of various patient populations.

In her own words Heather states, “Having a child with a rare disease opened my eyes to a world that I never knew existed, and I am dedicated to uncovering best practices, removing barriers, and spreading this knowledge to others impacted by this and other diseases. I am eager for an opportunity to contribute to the cystinosis/rare disease community working alongside other parents with a shared mentality and common goals around increasing education and advocacy.”

Terri lives in Novi, Michigan with her family and has been a member of the cystinosis community since her youngest son, Steve was diagnosed in 1989 at 18 months of age. She has found support and connection with families through CRN, and other cystinosis organizations through the years, attending many conferences. She has been the witness to ‘hope’ over the last 34 years as treatments, therapies, medications and an improved quality of life has been made possible through the efforts of so many. Terri is a past CRN board member (2011-2018) and has served as a past VP of Education & Awareness and Secretary. She is currently retired, but has a BSN from Indiana University, and has also worked as a paraeducator in special education for many years.

She looks forward to using her experience and passion for cystinosis to further CRN’s goals of family support, education, and promoting continued research.
ALAB Survey for Newsletter & Monthly Meet-up

Let’s Keep in Touch!

ALAB will be having meet-ups once a month.
Stay tuned on social media for dates and times!

Search “Adult Leadership Advisory Board”

@cystinosisalab1
@cystinosisTEENS

What do you need as an adult living with cystinosis? How can ALAB support you? We would love your feedback. Scan the QR code below.
CRN Bids Farewell to Christy Greeley

By Terri Schleuder, Director

For over 20 years Christy Greeley has been a ‘force of nature’ in the Cystinosis Research Network. She attended her first cystinosis conference in 2001 in Las Vegas shortly after her son, Jack, was diagnosed. Soon after she jumped in with both feet and has never looked back.

Serving in many capacities on the Board over the years as President, VP of Research and long time Executive Director of CRN, Christy’s leadership skills, organizational skills, her ability to connect with families, patients, doctors, researchers, industry partners, and many global advocacy groups has helped CRN to evolve into the premier Cystinosis advocacy organization it is today.

Thank you, is too small a word, but we do say THANK YOU. To say you will be missed is an understatement to be sure.

From your colleagues, and friends in the cystinosis community:

“Thank you, Christy, for encouraging me by your example to be the best advocate I could be for the cystinosis community, but more importantly, thank you for being my friend! I cherish the friendship we have and will be forever grateful to YOU for the respect this organization will continue to have in the rare disease world!”

- Marybeth Krummenacker

“I just met Christy for the first time in Nashville; however, she immediately welcomed me to the group as if we had known each other for years. Her knowledge is not only impressive, but speaks to a greater passion for the group and the disease as a whole. Parents like Christy are the reason those of us with younger children have a smoother path in disease management, and I’m beyond thankful for her contributions (past, present, and future) to cystinosis and its community.”

- Heather Rothrock

“Christy’s legacy is one of advocacy and knowing that this world we live in requires everyone to work together, to get where we want/need to be. Christy has shown me that this disease does not have to run your life, and you can do it all, but it doesn’t have to be done alone.”

- Chelsea Meschke

“From when we first met via NORD and continuing today to Professional Patient Advocates in Life Sciences (PPALS), I have admired and respected your tenacity, humor and positivity. You are indeed a force to be reckoned with (said with love)! Best wishes!”

- Jean Campbell

“Great leaders do not set out to be leaders…they set out to make a difference…it is never about the role…it is always about the goal…” The Cystinosis Research Network has thrived and made so many material contributions to the lives of individuals living with Cystinosis as a result of Christy’s leadership.

Christy Greeley, thank you for making a difference and your continuing efforts towards achieving CRN’s goals.”

- Jose Morales
I am always in awe of her ability to lead, guide and make our organization the best! She can see all.

- Karen Gledhill

I appreciate your helping me to make a smooth transfer and set up of the Deanna Lynn Potts Scholarship to CRN. I want to see young adults being fruitful and able to achieve their goals in life. I am glad I can honor my daughter in this way, thanks to your support.

- Gail Potts

Christy has been a beacon of light and leadership for our cystinosis community. In our short time since diagnosis, I have been so honored to spend time learning from her. Thank you for the inspiration, motivation, and encouragement you’ve given throughout the years to this community.

- Kristina Sevel

Christy has always brought her scientific expertise and her parent expertise very generously to the work of the Cystinosis Community Advisory Board. In Cystinosis Ireland we have valued the partnership we have built with CRN under her leadership. Personally she’s a dear friend that I always look forward to catching up with.

- Anne Marie O’Dowd

Christy has dedicated her spare time to helping our Cystinosis community out. Whether it be scholarships for financing, providing educational resources or putting together fun and educational filled conferences. She also started to give adults with Cystinosis more of a voice by starting Adult Leadership Advisory Board a few years ago.

- Jana Healy

Christy has been an inspiration to all of us as professionals representing her role as a mother of a child with cystinosis while exemplifying the importance of advocacy and funding for cystinosis research. It has been a privilege to have known and worked with Christy and I hope this will continue in an even larger role that she will now assume.

- Dr. Rick Kaskel

I have always been so impressed with her ability to connect with people and her relationships with our industry partners and medical professionals. She is a force to be reckoned with and her knowledge and leadership will be missed.

- Jen Wyman
CRN Bids Farewell to Christy Greeley, continued

“Life became more meaningful when I met Christy Greeley through CRN nearly 20 years ago right after my 2 young kids were diagnosed with Cystinosis. Her confidence and knowledge navigating life as mom, advocate and leader for Jack was infectious. We both were driven by better futures for our Cystinosis community and it gave us hope that we were doing something beyond ourselves.”

- Brittney LeBeau

“Dear Christy, your tireless efforts and dedication have made an incredible impact on countless individuals affected by cystinosis, and I am truly grateful for all you have accomplished. May this next phase for you and your family be filled with joy, fulfillment, and new adventures. I’m excited to see the wonderful things that lie ahead for you. Wishing you all the best.”

- Colleen Hammond

“I want to thank you, Christy, for your astute and dedicated leadership of the CRN, combining your scientific knowledge with humanism, organizational acumen, and managerial expertise. You made the Network thrive and left an indelible imprint on so many families and physicians! You have my personal and professional gratitude and admiration! All the very best.”

- Dr. Bill Gahl

“Respected, compassionate, tenacious, resourceful, mother, and friend. Those are the words that first come to mind when I think of Christy. She is an incredible person with a drive to better the lives of the many people living with cystinosis. It has truly been a blessing to call her my friend. With Respect and love.”

- Lesli King

Cystinosis Memorial Fund Recipient

By Karen Gledhill, Secretary

The Cystinosis Memorial Fund (CMF) was a fund established to help teens and young adults living with cystinosis. The goal is to instill added confidence and improve the skills and abilities of recipients in our community to flourish and fulfill their potential.

The fund will provide a stipend to help those living with cystinosis pay for a variety of educational activities. These include professional workshops and certifications, career and resume coaching, software programs, tutoring are a sample of technology focused services offered.

Our most recent community member receiving an award is Evan (also known as Ashe) LeBeau. They have been awarded funding to help with a nursing course as part of a phlebotomy program. They believe that being able to advocate for oneself, to be resilient and independent are some of the values that cystinosis has taught them. Congratulations!

The Cystinosis Memorial Fund
Learn more at cystinosis.org/cmf
As I begin to write this article for my Education & Awareness report for the next edition of our newsletter, I realize it has been one month since our conference in Nashville…and I am still in awe! This conference was extraordinary on so many levels! The immediate feeling on my arrival was…it was a big “family reunion…and it was! Seeing people for the first time, in person, in 4 years was overwhelming at times. It was such an honor to come back in person and have so many of our families together again and to meet so many new families as well! As I said in my session with Dr. Gahl and Dr. Thoene…things don’t just happen. It takes a lot of hard work and logistics and coordination to make it happen. This meeting and the conference planning team that put it together, should be so very proud of the collective work that was done. I know I am! We spent hours on conference calls and zoom calls coordinating every detail and continually fine tuning it along the way. Nothing we do as an organization is done alone. We work together and I think that was evident. We are a TEAM! Not only do we have the wonderful support of remarkable physicians and scientists, but also the support of our industry partners. That partnership enabled us to award scholarships to over 50 families…the most ever! We heard from our “pioneer,” Jordan Janz on his incredible journey as the first patient in the stem cell trial. Jordan was so gracious in speaking to our families and sharing his personal experience in his journey. This offered so much hope and promise for a brighter future! (Thanks to his mom, Barb, for reminding me of the hug I gave her at her first conference and the smiles she saw on the faces of a group of moms). Thank you to Jordan for paving the way. It sure looked like you had a great time!

I could not be prouder of the organization that IS Cystinosis Research Network. What began as a small, rare disease support group over 20 years ago, has grown into not only a well-respected advocacy organization within the cystinosis community, but also within the rare disease community as a whole. As I have been told by people in different rare disease communities …CRN is the gold standard of advocacy organizations. I am so proud of that! I am honored as well to serve beside some of the best people in the world on this Board of Directors, both past and present. I am excited to work with our new board as well. The feeling I came away with from this meeting is rejuvenation. I am excited for a better future for those who live with cystinosis every day! Even after 34 years of living with this disease … each day can be a challenge, but knowing you can reach out to anyone, at anytime …in any part of the world…is just incredible! Even though we all heard from the best and most brilliant experts on many levels, there is still nothing better than hearing from a family who lives with what you live with every single day. We are a blessed community in so many ways. We will continue with our presence at upcoming professional meetings. The next one is The American Society of Nephrology (ASN) meeting in Philadelphia in November. These meetings allow CRN to be represented, and to be a presence to meet and educate the nephrology community and physicians. We can tell “our” stories of what it is like to live with a rare disease like cystinosis. I have said for a long time-exhibiting
Education & Awareness Update, continued

with our CRN information is helping to educate the adult (and pediatric) nephrologists and physicians who are often treating our loved ones already. We also continue to network and communicate with other rare disease communities and umbrella organizations like NORD, Global Genes, Genetic Alliance, and Every Life Foundation. This allows us to share timely information and updates on important legislation, and also provides us the opportunity to attend and participate in their meetings, to be a voice for the cystinosis community. As we go to print, we will be announcing our 2023/24 Academic Scholarship recipients, which once again is an opportunity for our small community to take advantage of the generosity of all of our CRN donors. None of this could be done without your help and ongoing support.

Finally, I will leave you with this thought…we may be small, but we are mighty!! Thank you all for being a part of this mighty journey together! With GRATITUDE…OPTIMISM is SUSTAINABLE!

Academic Scholarship Recipients

By Gail Potts, Director

The CRN is happy to announce the 2023 recipients of the Individual with Cystinosis, and Deanna Lynn Potts Scholarship Awards.

The recipient of the Individual with Cystinosis award is Carina Jaramillo. Carina is attending DeVry University studying Medical Billing for certification. Carina has previously taken classes for certification in medical interpretation. She was encouraged to become a CMA and was certified in this field. Now she wants to advance her field in Medical Billing and Coding education.

We were pleased to award the Deanna Lynn Potts Scholarships this year, to Tina Flerchinger. Tina is attending Lewis-Clark State College, Lewiston, ID where she is majoring in Pre-Radiographic Science. Tina received high recommendations from her advisors for being a highly motivated and enthusiastic individual. Our sincere best wishes to all our amazing scholarship winners for a successful future!
Cystinosis Network Europe was delighted to be represented at the CRN conference in Nashville in July, with Denise Dunne presenting on the work of the organization in the last four years. She also gave an update on the Worldwide Cystinosis Community Advisory Board and its work. A summary of this work can be seen on the CAB poster.

CNE has also recently announced that the CNE International Conference will take place in July 2024 in Manchester, England. All are very welcome to attend and we will publish details as they are confirmed on the conference web page - https://www.cystinosis-europe.eu/our-conference.

CNE will hold our member update and AGM online on 25th September. For information on CNE, the CAB or the 2024 conference, please contact denise.dunne@cystinosis-europe.eu.

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**Worldwide Cystinosis Community Advisory Board**

**What are the key principles of the CAB?**

1. Neutral setting
2. Confidentiality
3. Transparency
4. Sharing
5. Openness
6. Optimisation of research & development
7. A joint action plan as an outcome of the meeting

**What is a Community Advisory Board?**

- A group of patient representatives who offer their expertise to public or private sponsors of clinical and other research
- The same group of representatives advises several sponsors in their field
- Agenda and secretariat driven by the patients
- The Cystinosis-CAB members are adults with cystinosis or parents or carers of people with cystinosis. All are experts in the condition and educated in research and research processes.
- Sponsors of research include pharmaceutical/healthcare companies; and researchers in hospitals, universities, and other settings.

**Who are the Worldwide Cystinosis Community Advisory Board?**

The CAB includes representatives from:
- AIRE España
- Asociación Cisticosis España
- Asociación Mexicana de Cisticosis AC
- Cystinosis Ireland (incl Northern Ireland)
- Cystinosis Foundation UK
- Cystinosis Research Network, USA
- Cystinose-Selbsthilfe e.V., Germany
- Dutch-Flemish Cystinosis Group
- Vaincre les Additioles Lysosomiales (VML) France

This group is nominated to represent our full membership – www.cystinosis-europe.eu

**What can the CAB offer?**

Patient representatives offer their expertise on:
- Patient outreach
- Education on research
- Clinical studies and their design
- Criteria for participation
- Informed consent forms and processes
- Compassionate use programmes
- Retention of participants
- Reporting on results
- Input and feedback on the development of tools for community use
- Patient input at the earliest stage of project development

**Worldwide Cystinosis CAB to date**

- July 2018: briefing from EURORDIS at Berlin Cystinosis Conference
- January 2019: first internal meeting and commitment to proceed; Chair elected.
- March 2019: training begins
- April 2019: discussions with Sponsors begin
- November 2019: first CAB and Sponsors meeting held in Dublin
- April & May 2021: virtual CAB and Sponsor meetings
- March 2023: In person CAB and Sponsor meetings
- Ongoing: training and discussion of issues of relevance and importance to the community and research priorities.
Cystinosis
Patient Assistance Programs

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.
Cystinosis
Patient Assistance Programs

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

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

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 RareCare™ 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.

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.
Annual PAS and ASN Kidney Week Update
The Importance of CRN’s Presence at 2 National Conferences

By Carol Hughes, Director

It is an honor to share with you information about our attendance at medical conferences with whom we exhibit. We always include wonderful photos of our booth. Now I’d like to take you behind the scenes to share the ‘nuts and bolts’ and the direct importance it is to our families & community. The two main conferences we attend are:

1. Pediatric Academic Societies, (PAS). Pediatric physicians who practice or research in various areas of medicine attend. Such as: nephrology, cardiology, endocrinology, gastroenterology, neurology and more. PAS is held in the spring, very close to Cystinosis Awareness Day, (CAD), May 7th. Events are held in alternating cities.

2. American Society of Nephrology (ASN) includes adult & pediatric nephrologists practicing, and in the research field as well. ASN Kidney Week is held in the fall annually in alternating cities.

Dr. Rick Kaskel, has specifically been the driving force that got us involved in this ‘exhibiting journey’ with his personal guidance and inspiration. He has been a member of our Scientific Review Committee since CRN’s inception and a huge proponent for us to advocate. His vision has always been for us to have exposure for our sweet, very small ‘rare disease community.’ What a blessing this has been for those living with cystinosis. We are able to participate annually at both the PAS and ASN Medical Conferences. At the very first exhibiting conference we arrived carrying a poster board with our kid’s pictures pasted haphazardly and a few brochures. We have come a long way in the last 25 years!

Many exciting relationships and connections are formed for the benefit of our children and adults who live with cystinosis. In addition, we provide useful literature, such as but not limited to:

- Brochures, parent handbooks, transition guides
- Zip drives loaded with cystinosis information and education

This is all available online at https://cystinosis.org.

Our booths also have giveaways including ever delicious mints and chocolate kisses! Most importantly, physicians share this knowledge with their patients and let them know that we are a voice for all the families, especially the newly diagnosed and those who may feel isolated. We can all relate to the feeling of isolation, at times, and the distress felt upon our diagnosis and throughout years of caregiving and the bumps that come along the way. It also gives us an opportunity to share our successes as well, which are many!

The conferences are also an opportunity for us to meet and educate doctors, medical students, and others in the scientific world. There are medical meetings and continuing education courses being held in separate breakout sessions simultaneously throughout the day as well as general group sessions.

In the convention center there are poster sessions on the outskirts of the booths that display the latest medical findings and results in many areas of research. It reminds me of our kids’ annual Science Fair, on an adult level. Most of our CRN Scientific and Medical Advisory Board Members have displayed posters in these sessions over the years pertaining to cystinosis and transplants as well. The booths themselves, (mostly 10ft x 10ft in size), are comprised of hospitals, medical literature companies, pharmaceuticals, and a very small group of non-profit organizations, such as CRN.

Exhibiting at these events gives us the opportunity to meet with a broad
Annual PAS and ASN Kidney Week Update, continued

similar journeys as the CRN.
The opportunity to exhibit is beyond rewarding and worth the time and effort invested. If just one new diagnosis is obtained from our participation, the effort to exhibit is a success! The generosity of donors makes attendance with a CRN booth possible. I am excited for the upcoming American Society of Nephrology (ASN) Kidney Week in November in Philadelphia!

Thank you for letting me share our ‘nuts and bolts’ of what it takes to exhibit at these professional medical conferences and the importance of advocating for the betterment of the cystinosis community on a level that most are unaware of. Hope it enlightens and inspires you in some small way to be a voice for your loved one (or yourself).

spectrum of attendees on a personal level. We get to meet with physicians, residents in training, pharmacists, and medical students deciding in what field to practice.

ASN and PAS conferences last for several days and are attended by thousands. CRN Board Members volunteer their time. It is both a difficult and rewarding encounter at the same time. We share our own experiences with attendees on a personal level. I’ve had very touching moments meeting the most amazing doctors and other health care professionals over the years that will forever stay in my heart. We get to greet new faces from all over the world as well as familiar faces - one is my neighbor who is a nephrologist - small world! Hi Ana! We share helpful hints and resources and support from other small non-profit organizations on

For ways to participate, email info@cystinosis.org
Dear Cystinosis Community,

I hope this letter finds you well and filled with the same sense of gratitude and excitement that I’m experiencing as I write to you today. The past year has been a remarkable journey for all of us at the Cystinosis Research Network, and I’m thrilled to share our progress and achievements with you.

This July marked a pivotal moment as we gathered together for our first in-person family conference since 2019, in the vibrant city of Nashville, Tennessee. The joy of reuniting with familiar faces and welcoming new ones was palpable, reminding us all, of the strength and unity that defines our community. The conference brought a renewed sense of purpose, invigorating our commitment to advancing research, supporting families, and raising awareness about cystinosis.

One of the pillars that make our progress possible is the remarkable support we receive from our industry sponsors. This year’s conference was made even more memorable thanks to their unwavering dedication. Our industry sponsors truly embody the spirit of collaboration and innovation. Without the generosity of Horizon Therapeutics, Leadiant Bioscience, and Recordati Rare Disease, none of these potential ventures would be possible. We have grossed over $400,000 in direct funding via an array of targeted and open-ended grants in 2023, and look to eclipse this number in 2024.

As we reflect on our accomplishments, we cannot overlook the invaluable contributions of our Cystinosis Warriors Impact Program. This program has been a source of inspiration, uniting individuals affected by cystinosis and their families. Your stories, resilience, and determination continue to drive our mission forward, and we are honored to stand alongside you on this journey.

In line with our commitment to accessibility and support, we’re thrilled to share our continued collaboration with the PCs for People program. Providing free computers and internet access to families in need is a small step towards easing the challenges that can come with managing cystinosis. 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.

Bromberg & Associates maintains its commitment to providing CRN translation and telephonic interpretation services for Spanish-speaking individuals and families. Furthermore, the company has extended its global outreach to encompass cystinosis patients worldwide, now delivering translation solutions across more than 200 diverse languages, and this catalog is continuously growing.

Have you or someone you care about recently received a diagnosis? Are you moving towards the transplantation phase or transitioning to an adult primary care provider?

Explore the remarkable offerings of our Care Package Program, designed to provide assistance to individuals and families dealing with cystinosis during challenging periods. We’re here to send you a customized package containing informative resources and helpful resources tailored to your specific stage in the cystinosis experience. To enroll in a free package, visit https://cystinosis.org/care-package/ and complete a short form.

In the spirit of giving back, I invite you to explore the opportunities to give that are close to our hearts. Be on the lookout for Giving Tuesday this November 28th; a wonderful moment to make a difference, and Cystinosis Awareness Day next May 7th which provides a platform to raise awareness and show your support.

Thank you for being an integral part of the Cystinosis Research Network. Your support, determination, and passion drive our progress and inspire us to reach greater heights. Together, we are making strides towards a brighter future for individuals and families affected by cystinosis.

Jonathan Dicks with Jen Wyman and Christy Greeley at the Nashville conference
We are committed
to providing personalized services
to help patients get the most
out of their therapies.

Learn more about the resources available at www.CYSTADROPS.com
I had the privilege of participating in the three-night Raregivers Men’s Retreat recently organized by AngelAid—an incredible organization dedicated to supporting families navigating the complexities of rare diseases. The retreat, nestled against the serene backdrop of the rugged Santa Ana Mountains, unfolded from Thursday, June 8th, marking the commencement of a transformative experience for attending rare caregivers—specifically fathers of sick or deceased rare children, as well as rare patients themselves. The retreat aimed to offer these men a space of solace, healing, and strength among fellow dads and patients who share parallel experiences.

Throughout the three nights, a meticulously designed program catered to the distinct emotional and psychological needs of these extraordinary individuals. Central to this retreat were focused therapy sessions, expertly guided by psychologists and counselors who skillfully navigated us through our grief and complex emotions associated with profound loss. These sessions provided a supportive environment to share stories, genuinely express feelings, and receive empathetic guidance from professionals well-versed in the realm of rare diseases.

The retreat also featured workshops and group discussions, fostering connections and forming a tightly-knit support network. Through open dialogues, we found solace in the knowledge that our personal struggles were shared, forging deep connections and lasting friendships that I believe will extend far beyond the retreat itself.

In addition to therapeutic sessions, the retreat offered a variety of activities promoting self-care and emotional well-being. Guided meditation, sound baths, and yoga provided tranquil moments for finding “stillness” and rejuvenation. Outdoor pursuits like hiking and group fitness sessions encouraged camaraderie, challenging us physically and boosting confidence and self-esteem.

Meals were shared in a communal setting, becoming opportunities for bonding over shared journeys, and exchanging stories and insights. Thoughtfully prepared culinary offerings took into consideration dietary restrictions and preferences, ensuring nourishment on both physical and emotional levels. Experiencing the profound impact of this retreat for the first time was both humbling and inspiring. The Raregivers Men’s Retreat provided a sanctuary where grief could be freely expressed, healing found, and strength gathered for the journey ahead. It stood as a testament to the potency of community, empathy, and compassion.

This retreat not only equipped us with tools to navigate grief and guilt, but also empowered us to honor the memories of our rare loved ones and embrace life with hope and resilience. To the group of brothers who attended, may the connections formed, the healing encountered, and newfound strength propel you forward on your journey. And to AngelAid, a heartfelt thank you for orchestrating such an exceptional event—a source of solace and support for these exceptional men who have faced unimaginable challenges with grace and courage.

By Jonathan Dicks, Vice President of Development
Cystinosis Ireland marked our 20th anniversary with a family and scientific meeting held in April in Barretstown, the Irish edition of the Paul Newman Hole in the Wall Gang Camps. The meeting was an incredible success with more than 100 family members, scientists and clinicians coming together to learn and share. The 9th Dublin Cystinosis Workshop was held as part of the weekend and this year’s themes were: 1) Evolution in mechanistic / pathogenic insights in cystinosis, 2) Innovative treatment approaches/ Innovation in treatment and 3) Novel clinical aspects. The Workshop heard from researchers and clinicians from the USA, Canada, New Zealand, UK and Europe (including Ireland) who shared insights and most recent findings as efforts continue to identify new drug targets and effective therapies for cystinosis and ultimately to find a cure for this ultra-rare disease. As part of our poster presentation, Louise Medaer of Katholieke Universiteit Leuven, was awarded the Professor Roz Anderson Memorial Prize for her presentation of her work on S Isogenic human cell models to unravel the underlying mechanism of cystinosis myopathy. The Family Workshop was fun-filled and action packed. Adult patients, parents and caregivers were given the opportunity to engage with our research and clinical colleagues to learn more about areas of importance to the community including therapy development, new areas of research and potential treatments of the future. Our families also engaged with our research community to discuss current research projects and gain a greater understanding of the research process. Our up and coming researchers were challenged to explain their work to a lay audience, who voted on the best presentation. The Cystinosis Ireland Award for Best Oral Presentation to a Non Scientific Audience was awarded to Hayley Chang of the Frederick J. and Marion A Schindler Cognitive Neurophysiology Lab (CNL), University of Rochester School of Medicine and Dentistry, New York, for her work on Development and Characterization of a Cystinosis Knock-Out Mouse Model Using CRISPR/Cas9.

While the adults were learning and sharing their perspective with the scientific attendees, the children were enjoying activities including archery, fishing, canoeing, wall climbing, crazy golf, movie making and prank parties. The sun shone and screens were abandoned for an opportunity to make new friends and try new experiences. The main feedback from the weekend was “when will we be coming back”!

For more information about Cystinosis Ireland and our work, please contact denise.dunne@cystinosis.ie
Have you attended an impact program yet?

The IMPACT Program is specifically designed for people living with cystinosis and their families, and sponsored by Horizon Therapeutics.

- Learn about living with cystinosis
- Connect with others impacted by this condition—hear and share ideas, feelings, and helpful advice
- Learn about medication:
  - the critical importance of continuous cystine control through cystine-depleting therapy (CDT)
  - PROCYSBI (cysteamine bitartrate) delayed-release capsules and delayed-release oral granules for the treatment of nephropathic cystinosis
- Meetings will occur in both the virtual (online) setting and live, in person

Even if you’ve attended an IMPACT Program previously, please check back in with us!
The IMPACT Program has new topics created specifically for different cystinosis patient experiences—adults, teens, and families/caregivers of people living with cystinosis.

To RSVP to one of these meetings or to find out about programs occurring near you, please call 602-953-2552 or visit www.procysbi.com/Cost-Savings-and-Support

Use and Important Safety Information

What is the most important safety information I should know about PROCYSBI?

PROCYSBI can cause serious side effects, including:

- **Skin, bone, and joint problems.** People treated with high doses of cysteamine bitartrate may develop abnormal changes of their skin and bones, such as stretch marks, bone injuries (such as fractures), bone deformities, and joint problems. Check your skin while taking PROCYSBI. Tell your doctor if you notice any skin changes or problems with your bones or joints. Your doctor will check you for these problems.

- **Skin rash.** Skin rash is common with cysteamine bitartrate and may sometimes be severe. **Tell your doctor right away if you get a skin rash.** Your dose of PROCYSBI may need to be decreased until the rash goes away. If the rash is severe, your doctor may tell you to stop taking PROCYSBI.

Please see additional Important Safety Information on the next page, and visit www.hzndocs.com/PROCYSBI-Patient-Information.pdf for the Patient Package Insert.
IMPORTANT SAFETY INFORMATION (continued)

- **Stomach and bowel (intestinal) problems.** Some people who take other medicines that contain cysteamine bitartrate may develop ulcers and bleeding in their stomach or bowel. People treated with PROCYSBI may also develop abnormal swelling and narrowing of the large bowel which must be treated promptly. **Tell your doctor right away** if you get abdominal pain, bloody or persistent diarrhea, bloating, nausea, vomiting, loss of appetite, vomit blood, poor weight gain or weight loss.

- **Central nervous system symptoms.** Some people who take other medicines that contain cysteamine bitartrate develop seizures, depression, and become very sleepy. The medicine may affect how your brain is working (encephalopathy). **Tell your doctor right away** if you develop any of these symptoms.

- **Low white blood cell count and certain abnormal liver function blood tests.** Your doctor should check you for these problems.

- **Benign intracranial hypertension** (pseudotumor cerebri) has happened in some people who take immediate-release cysteamine bitartrate. This is a condition where there is high pressure in the fluid around the brain. Your doctor should do eye examinations to find and treat this problem early. **Tell your doctor right away if you develop any of the following symptoms while taking PROCYSBI:** headache, buzzing or “whooshing” sound in the ear, dizziness, nausea, double vision, blurry vision, loss of vision, pain behind the eye, or pain with eye movement.

**What is PROCYSBI?**

PROCYSBI (cysteamine bitartrate) delayed-release capsules and delayed-release oral granules is a prescription medicine used to treat nephropathic cystinosis in adults and children 1 year of age and older. It is not known if PROCYSBI is safe and effective in children under 1 year of age.

Do not take PROCYSBI if you are allergic to penicillamine or cysteamine.

Before taking PROCYSBI, tell your doctor about all your medical conditions, including if you:
- drink alcohol.
- have a skin rash or bone problems.
- have or have had stomach or bowel (intestinal) problems including ulcers or bleeding.

- have a history of seizures, lack of energy, unusual sleepiness, depression, or changes in your ability to think clearly.
- have liver or blood problems.
- are pregnant or plan to become pregnant. It is not known if PROCYSBI will harm your unborn baby. Tell your doctor right away if you think that you are pregnant. Talk with your doctor about the benefits and risks of taking PROCYSBI during pregnancy
- are breastfeeding or plan to breastfeed. You should not breastfeed during treatment with PROCYSBI. Talk with your doctor about the best way to feed your baby if you take PROCYSBI.

Tell your doctor about all the medicines you take, including prescription and over the counter medicines, vitamins, dietary and herbal supplements. Know the medicines you take. Keep a list of them to show your doctor and pharmacist when you get a new medicine.

**What should I avoid while taking PROCYSBI?**

- Do not drive or operate machinery until you know how PROCYSBI affects you. PROCYSBI can make you sleepy or less alert than normal.
- Do not drink alcohol if you take PROCYSBI. Drinking alcohol while taking PROCYSBI may change how PROCYSBI works and may cause an increase in the amount of PROCYSBI in your blood that may cause serious side effects.

**What are the possible side effects of PROCYSBI?**

PROCYSBI can cause serious side effects, including:
- See “What is the most important information I should know about PROCYSBI?”

The most common side effects of PROCYSBI include:
- vomiting, nausea, stomach (abdominal) pain, pink eye, diarrhea, cold, tiredness, flu, headache, problems with body salts or electrolytes, infection of ear, nose or throat, joint pain.

These are not all the possible side effects of PROCYSBI. Call your doctor for medical information about side effects. You may report side effects to FDA at 1-800-FDA-1088.

For additional important safety information, please visit www.hzndocs.com/PROCYSBI-Patient-Information.pdf for the Patient Package Insert, and discuss with your doctor.
Cystinosis Family Day 2023, Acapulco

By Victor Gomez

Once again Acapulco Mexico was the venue for the 3rd Cystinosis Family Day 2023 Meeting, 12 families were present to enjoy a weekend full of fun, motivation, and learning.

CONFERENCE HIGHLIGHTS

Psychology
Maricela Lopez Roa and Jessica Marcela Villeda Serna, a team of psychologists were in charge of guiding the group with a series of dynamics which helped to break the ice and reinforce the support the patient needs from the whole family.

Ophthalmology
Dr. Liang Hong, an ophthalmologist attached to the Hospital of Ophthalmology uninez-Vingts National Paris, gave a lecture via zoom in which she highlighted:
- How the ophthalmological management of patients with Cystinosis is given in Paris
- Studies demonstrating the effectiveness of Cystadrops treatment
- Treatment management tips and recommended doses in children and adults
- Consequences of not using ophthalmic therapy

Nephrology
Dr. Juan Guillermo Cardenas Aguilar, Pediatric Nephrologist of the Universidad Militar Nueva Granada Bogotá Colombia, spoke about specific data on Cystinosis:
- Clinical data in Cystinosis
- What are the recommended values in a pediatric patient with Cystinosis?
- Specific comprehensive therapy for Cystinosis
- Technological tools in the management of a patient with Cystinosis (app)

Patient Story
Irati Vilariño patient with cystinosis living in Spain & member Cystinosis Organization Spain spoke to the audience about her experience living with cystinosis, and personal living, work, hobbies and hard work collaborating in Cystinosis Organization Spain. She motivated the patients to live life normally, work, achieve goals besides cystinosis.

Nutrition
Estefania Zuluaga Nutritionist affiliated to the Colombian Association of Inborn Errors of Metabolism (ACPEIM) highlighted outstanding information on nutrition for Cystinosis:
Cystinosis Family Day 2023, Acapulco, continued

- Nutrition in transplanted patients with Cystinosis
- Tips on healthy nutrition in Cystinosis
- Recommended values to balance a healthy diet in patients with Cystinosis.
- Experience in consultations with patients in Colombia

Cystinosis Organization Mexico

Finally, Victor Gomez, President of Asociación Mexicana de Cistinosis AC gave a report on the work of the association:

- Due to the problem of shortage by health sector institutions in Mexico, the association has donated immunosuppression treatment to patients.
- Access to cystine level analysis has been achieved, and we are currently working in collaboration with a Medical University to formalize a cystine analysis center.

- We are working hard to continue with medical training for ophthalmologists and nephrologists in the health sector.
- The association’s board of directors is renewed and a new Facebook channel for Mexican patients is being promoted, which will be used for support and guidance.

Finally, on Saturday night, a farewell party was organized to close the event.

Irati Vilariño From Spain
My name is Erica Hall, and my husband Andrew and I are blessed to be the parents of Marley, a two-year-old Cystinosis Warrior. We also have a baby girl due in January 2024, who is a carrier but does not have Cystinosis. Like most of our fellow Floridians, Andrew and I are both “from somewhere else.” I am originally from Connecticut, and he is originally from Ohio, but we have fallen in love with Florida life.

Our journey to becoming parents is certainly not what we expected. After a miscarriage followed by a nerve-wracking bedrest pregnancy with Marley, we were thrilled when she arrived perfectly healthy and right on time in February 2021.

Marley was a phenomenal eater from the moment she was born. She was a dream baby, sleeping through the night at 8 weeks old, growing, hitting milestones perfectly, and charming everyone with her easy-going demeanor. At 5 months old, the pediatrician mentioned that Marley had fallen off her growth curve. Despite supplementing with formula, she remained in the 7th percentile, and we just assumed it was because I am also petite. At 9 months we began switching completely to formula and that was the beginning of the end of normalcy for us. She became chronically constipated no matter what we did or what formula we tried. She was constantly thirsty, peeing frequently, and no longer growing at all. Her eating gradually decreased until she stopped completely. I took her to gastroenterologists, allergists, nutritionists, feeding therapists, natural doctors, and everything I could think of, but no one could find anything wrong with Marley. Finally, a bout with Covid at 15 months old sent Marley to the hospital where we would spend 3 agonizing weeks isolated in the PICU where her Fanconi Syndrome was discovered. A team of nephrologists told us they suspected Cystinosis. The first year post diagnosis, was very difficult, as we tried lots of different medications trying to get Marley’s labs to be stable and adjust to our new life as medical parents. We began to hit a bit of a stride in November after switching to Cystagon and switching her from formula feeds to blended foods.

Today Marley seems almost as normal as any of her peers except the noticeable difference that she doesn’t eat orally. She is a chunky toddler with plenty of ‘2-year-old attitude’ to spare. She is very intelligent and talks constantly. She is currently loving all things superhero, outer space, and dinosaurs! Our “next steps” are to put her on a pump for hydration overnight, increase her Cystagon to the next dosage, and get her PTH levels down a bit more. And hopefully someday soon, get her to eat again!

We hope and pray for a cure for Cystinosis so our sweet girl can truly thrive!
Virtual camp provides a break from dialysis, hospitals and challenges — enriching the lives of children with kidney disease by allowing them to just be kids. Share life-experiences, form friendships, socialize with peers and engage in creative, constructive virtual activities led by dedicated AKF staff.

Learn more and register today at www.kidneyfund.org/national-virtual-cystinosis-camp

Questions? Contact Lianna Chase at lchase@kidneyfund.org
I am so excited to be a part of the CRN Board as the new Vice President of Family Support. My family became a part of this beautiful Cystinosis community 6 years ago after our oldest son Jaxon was diagnosed. Our youngest Myles also has cystinosis and was diagnosed at 3 weeks. In those years we have cried, been angry beyond belief, laughed, and learned our “new normal”. We have found such comfort in this community and know how much it means to have that support. Learning tricks, tips, and ways to navigate Cystinosis from those before us, and next to us has gotten our family through many difficult times. I hope to provide that same love, friendship, knowledge, and sense of family to those needing it. Sometimes asking for help can be a difficult task, but the CRN family is always here with a hand to hold, a shoulder to cry on, or something to make you laugh when you need it most. If you have any suggestions for topics to discuss, ways to increase our family network, or need to talk please feel free to reach out to myself at calto1cl@gmail.com or Jen Wyman at jenwyman@comcast.net.
In Memoriam

Channing Leigh-Ann O’Halloran

By Lanna Smith

Channing Leigh-Anne O’Halloran was diagnosed at 11 months old with Cystinosis in 2003 at Arnold Palmer Hospital in Orlando, FL. In 2004, her family moved to Gainesville, FL for her care at UF Health/ Shands. She also saw Dr. William Gahl at NIH every 2 years. She was featured in NIH Medline Plus magazine twice for the amazing strides she made and obstacles she overcame with Cystinosis. Throughout her life battling Cystinosis, she didn’t let it stop her from living her life to the fullest.

Channing’s passion was acting, and since age 10, she performed in professional theater, TV and film. She was in seasons 1 and 2 of “Stranger Things” on Netflix. She was also a cheerleader in school and excelled academically. Channing had a kidney transplant at age 17 in 2020. She was in good health and pursuing her dreams.

Channing Leigh-Ann was tragically taken on March 20th in a fatal car accident in which she was the passenger. She was 20 years old. On March 25th she was given an honor walk as she donated her heart, liver and pancreas. She was adored and cherished by her family. She was a true star and totally unforgettable.

Channing’s infectious personality, full of humor, charm and strength, inspired everyone who had the pleasure of knowing her and left an impact. She will be remembered as a beautiful warrior and always shined like a ray of sunshine. She leaves behind her mom and dad, Lanna Smith and James Stewart, and her 4 year old baby brother, Reef Jameson Stewart.

Kevin McCalla

By John and Susie McCalla

Kevin Patrick McCalla was born on July 27, 1984, in San Diego, CA. His parents, John and Susie, were both in the United States Marine Corps, so Kevin moved around quite a bit. The family had moved from California to Virginia when Kevin was less than one year old. During the long drive across the country, Kevin was constantly thirsty, sucking on a wet washcloth for much of the trip. Upon arrival in Quantico, VA., he was diagnosed with Fanconi Syndrome at the local Naval clinic. Further evaluation was conducted at the Bethesda Naval Hospital to determine the cause.

After a very long week of test after test, a blood sample was taken to the nearby National Institutes of Health by one of the interns on his personal moped. Soon after that John, Susie, and Kevin met Dr. Bill Gahl for the first time. Kevin was 13 months of age and immediately placed on the experimental form of cysteamine.

The McCalla family’s next assignment sent them to Pennsylvania and then on to North Carolina. Those
In Memoriam

Kevin McCalla, continued

locations were within reasonable distance to Maryland for needed follow-up visits. Trips to NIH also included the experimental protocol for cysteamine eye drops, which required drops every hour. The results of that experiment proved that these eye drops were effective. Many years later Kevin would recall the relationship he had with Ernie who took many photographs of Kevin’s eyes. Kevin also enjoyed the playroom at 9 West and the many art projects he worked on.

The next duty station for John, Susie, Michael, Kevin, and Kelly made appointments to the NIH a bit of a challenge. They were sent to Hawaii for the next four years. The entire family loved those years in Hawaii. When John retired from active duty after that tour, the family settled down in the Kerrville, Texas area. Kevin was in the 5th grade, his first year there, making many friends, loving art, and playing football. He graduated from Center Point High School and went on to receive a Bachelor of Arts in Studio Art from the University of Texas at Austin.

As a result of cystinosis, Kevin’s worsening kidney function necessitated two separate kidney transplants, both of which eventually failed. After three years of dialysis and many health issues, Kevin died of complications from cystinosis, on January 31, 2023.

Kevin’s love of art and storytelling led to the creation of a series of three comic books to both entertain and educate the cystinosis community.

To learn more about Kevin and his amazing cystinosis themed comic books check out this link:

Pam Woodward

By Frankie McGinnis

Long time cystinosis community member, former Board member, a loving mom, wife and grandma, a “cysta” and friend to all who knew her passed away after a brief battle with cancer December 14, 2022. This tribute to her was written by her dear friend, Frankie, shortly after she left us:

I have known for the last week and a half since I left Utah that today was coming. Every morning I have been hesitant to look at my phone as if...
I could somehow avoid what was looming. This morning the news came that you went to the next realm and your pain had ended.

When my gal was diagnosed with cystinosis my world was made bigger by all the lovely people who would become family as we fought for our children’s lives. The “cystas” I would gain would hold my hand figuratively and physically through many laughs and many tears. Pam and I quickly became close and have shared so much. Through all our many trips, conferences, fundraisers, escapes from life’s horrors our bond was set.

When I got word of her diagnosis, I knew I had to get to her, to have a moment. Plans were made and then things began progressing quicker than anticipated so like always I punted. To say I am grateful that I got to spend a couple of days lying in bed listening to her stories again is an understatement. At times it was like she was infusing me with the history of our life so that it could carry on. I am horrible with dates so that skill will go with her. My beautiful, precious ‘cysta’ told me when the doc asked her how she kept going when cancer was invading her body her answer was simple, she was tough.

So, to my Utes loving, f bomb dropping, “oh my heck” saying, keeper of all the memories my heart will miss you here. I have no doubt you are organizing Dude Cup 2.0 and loving on your precious Sierra and all the way too many loves that rejoiced when they got you in the next realm.

For those still here, your beloved Rock (who was literally your rock), Tahnie, Sookie, your brothers and the slew of people who love you I pray for peace. Until I see you again, this ‘cysta’ will miss you.
Could you walk us through how it felt to learn the kidney from the first transplant “fell asleep” or was not working properly?

Victor: It felt frustrating to know that after all my dad did to give me his kidney, it turned out not to work at all. It was very disheartening for me and my family as this was the first transplant and it turned out not to want to work no matter what we did to help it. I felt depressed after so many attempts to try and get this kidney to work properly and was on dialysis for a year and a half. It was hard for my family to even comprehend why it didn’t work and we are still trying to figure out what happened with it.

India: When I first found out that Victor needed another transplant, I was very surprised but ever since I was little, I told my parents I wanted to be the one that saved Victor and I knew that eventually, I would have to make the decision to officially start being tested and start the journey of being his next donor. At this point when we found out I was not already tested. My parents didn’t want me to have to worry about being his donor, especially since I was in school. My parents wanted me to be a backup plan for him when he was way older, but it happened sooner than they would have liked.

What were your thoughts when you found out your sister, India, was a match?

Victor: I was elated to know that my sister would be giving up one of her organs for my sake. I was extremely happy to realize that I would finally be
able to have a life outside of dialysis.

India: I knew that I was going to donate to Victor since I could comprehend that Victor had cystinosis. I saw how dialysis was taking a negative toll on his body, mental state, and overall life. I refused to watch my brother suffer more than he needed to. My family and friends were very supportive of my decision. Some visited me in the hospital, and it really made me feel good.

Please describe the process of preparing for your kidney transplant.

Victor: The process for preparing for transplant was a lengthy one to say the least. A lot of lab work to be done as well as many other tests that needed to be completed such as an ultrasound, an EKG, etc.

India, how did your medical team and family support and guide you?

India: My medical team was very supportive, and they made me feel like I had a safe space with them and that I could talk to them about anything. My kidney coordinator would check up on me frequently just to make sure that I was okay and if I had any questions, they answered them without hesitation. They made something I was very nervous about a comfortable experience.

My parents are our rock. They tagged our appointments, but I must give it to my mom because days when my dad wasn’t there, she was running back and forth from each side of the hospital to make sure that she wasn’t missing anything. My mom asked so many questions because, at the end of the day, this was Victor’s second transplant, and this time it wouldn’t be my dad on the table, it was me and that was terrifying for her because no one knew how it was going to go. But Dr. Wee reassured us that we were going to be okay in his hands and I don’t know where Victor would’ve been without him.

How were you feeling before surgery?

Victor: I’m not going to lie, I was a bit nervous before surgery seeing as what happened with the last one, but luckily we had the best surgeon in the kidney transplant department doing the surgery so that made me less uneasy and ready for surgery.

India: I was so nervous, I never had any big surgeries before so the entire process leading up to it was nerve-wracking. I know I should’ve voiced my feelings more, but I didn’t and before the surgery, the only thing on my mind was making sure that I wasn’t Victor’s next failed transplant. I wanted more than anything for him to be okay.

Victor, how was the procedure? And how it compare to your first transplant?

Victor: The procedure went by smoothly and without any problems for me. My mental and physical state of mind after this transplant was much better than the first one. I was able to move around and recovering better.

How was your surgery, India? How were you feeling physically and mentally afterward?

India: The surgery went well. Victor got discharged before me and was feeling great. The day after the surgery I, on the other hand, wasn’t doing as well. They switched my diet quicker than my stomach would have preferred and on top of my stomach pain my body was against me. The surgery was on a Friday and that Saturday was filled with a lot of vomiting and pain. When I was finally able to be discharged it was two days later and I was doing way better. I did have issues with my blood pressure

The Gift of Life, continued
being tacky but that went away rather quickly. After the surgery my mental state was good, I knew I did a good thing because Victor’s levels were already going down. It took me a while to get used to the four surgery scars but any type of negative thoughts I placed on my body quickly went away. Those scars display a moment of selflessness and love I have for Victor and I would never wish them away.

When you reflect on your sister’s decision to donate her kidney, how does it make you feel?

Victor: It makes me feel really happy that my sister made this tough decision for my sake. I will be forever grateful that she did this wonderful thing even though she didn’t have to.

Organ donors are often referred to as heroes. India, how do you feel about that title?

India: I may be seen as a hero but for me, I just wanted to save him. Victor is 23 years old now and before the surgery, he wasn’t in a good place. He was stuck and I wanted to give him a better life and that doesn’t make me a hero, it makes his keeper, his little sister, and I would do it all over again just to watch him get wheeled into my room with that big goofy grin of his.

Post-transplant, did your relationship with your sibling change?

Victor: The relationship we have as siblings didn’t really change as before the transplant we were already in a good relationship with one another.

India: Victor and I are the best of buds. We always have been, and I thank my parents for that. I would like to say that Victor took my sweet tooth with that transplant, and I haven’t wanted sweets since, so watching him eat chocolate and sweets makes everything even better.

How has your perspective on life been impacted by this experience?

Victor: After being on dialysis for over a year, my life revolved around just that and nothing else. Now that I’ve had this transplant and everything has been working properly, my life is back to how I want it to be. I am able to do things I want to do without dialysis getting in the way.

India: This experience grounded me. The feeling I had when I found out that I helped him is something I could never forget. This experience gave me a better outlook on life because my one decision to get tested evolved into me turning his life around completely and I thank God for blessing me with the ability to be his match.

How is your health and overall well-being today? Any long-term adjustments you’ve had to make?

Victor: Great so far. Everything is working just like it should with no issues to deal with. I’ve no longer had to take all my electrolytes anymore and my med list has been diminished greatly.

India: I am doing so well, I haven’t had any issues with my kidney function or anything else and the only thing I had to really adjust to was not being able to take ibuprofen, which ultimately sucks but I can low dose myself if it’s needed.

What advice would you offer to someone considering organ donation? Are there any misconceptions you’d like to clear up?

Victor: It may be a long process to deal with, but it will all be worth it in the end and the person that you are donating an organ to will greatly appreciate the gesture and sacrifice that you have made for them.

India: I had my surgery at Cleveland Clinic and they make the most kidney donations each year equaling more than 200 transplants so if you ever want to get information or even start getting tested talk to them; their team is the best in Cleveland (yes, I’m biased). The process isn’t hard, it isn’t scary, and the people make you feel comfortable and safe. If you ever feel like this is something you could do, take that leap and do it because knowing that you changed somebody’s life is the best reward you could ever ask for.
A Crystal-Clear Solution, in an Easy-To-Use Bottle

To treat corneal crystals in patients with cystinosis.
Please visit www.cystaran.com to learn more

What is CYSTARAN®?
CYSTARAN (cysteamine ophthalmic solution) 0.44% is an eyedrop medication used to treat cystine crystal accumulation in the corneas of patients who have cystinosis.

What is the most important information I should know about CYSTARAN?
- To help prevent contamination of the dropper tip and eyedrop medication, try to make sure that CYSTARAN is dropped directly onto the eye without touching it. Try not to touch the eyelids or surrounding areas with the dropper tip of the bottle when you are using CYSTARAN. Keep the bottle tightly closed when not in use.
- CYSTARAN contains an ingredient called benzalkonium chloride which can be absorbed by soft contact lenses. Remove contact lenses before using CYSTARAN eyedrops and wait at least 15 minutes before reinserting them.
- CYSTARAN should only be used as an eyedrop medication.

What are the side effects of CYSTARAN?
- The most common side effects of CYSTARAN, which have occurred in at least 10% of people using the medication, were sensitivity to light, eye redness, eye pain and irritation, and headache.

The risk information provided here is not comprehensive. To learn more, talk to your healthcare provider or pharmacist about CYSTARAN. The full FDA-approved product labeling can be found at www.cystaran.com.

You are encouraged to report negative side effects of prescription drugs to the FDA. Visit www.fda.gov/medwatch or call FDA at 1-800-FDA-1088.

For information about ordering CYSTARAN®, please contact our partner, AllianceRx Walgreens Pharmacy:
PHONE: 1-877-534-9627

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Live Like Laura Fun Fund Recipients Share their Gratitude

by Frankie McGinnis

The Live Like Laura Fund, in memory of Laura McGinnis, was started to allow those who live with cystinosis to experience adventures and fun as Laura loved to do. Below Gracie and Karolis, express what receiving the fund meant to them.

Learn more at cystinosis.org/llff

I am so grateful to say that I received the live like Laura fund for my 21st birthday. I had the opportunity of meeting Laura a handful of times in person which were always full of laughter as far as I remember. We were alike in a lot of ways other than cystinosis. We followed each other on social media. I remember seeing Laura post pictures of herself on all of these trips and I couldn’t wait to graduate and move out so I could do the same. She showed me even with cystinosis, I can. My 21st birthday was such a fun-filled weekend. I went to Atlanta (where I currently live, a year later with my fabulous roommate I met that weekend.) I went to my 1st drag show, Top Golf, Dave and Busters, did some shopping, and ate so much amazing food. I also got the pleasure of having a little chat with our cysta, Mrs. Willow Pill, and taking her fave drink as my first legal drink (it’s a shot of tequila with lime, of course). That weekend was spent making memories, laughing, meeting new people, and so much love. Thank you so much for this award, it was well used, and it was all with Laura in mind.

- Gracie Smith

I wished for an electric scooter and was so happy when my wish was granted. My name is Karolis and I live outside Uppsala in Sweden with my mum, little brother and two cats. I was diagnosed with Cystinosis when I was 4.5 years old and I now have chronic kidney disease, and will soon need a transplant. I am very tired and my leg hurts from muscle wasting. I wished for an electric scooter and was so happy when my wish was granted. The scooter has helped me get to school by myself instead of mum having to drive me and it has also made it possible to hang with my friends when they are out with their bikes and I could not keep up with their speed.

- Karolis Schröder
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 the patient or family member has been diagnosed with any of the following conditions, they qualify for document translation and interpreting services by telephone provided by the **Bromberg & Associates** team at no-cost:

- Rare metabolic conditions
- Rare kidney disease
- Uncontrolled Gout
- Primary Immune Deficiencies
- Graves and/or Thyroid Eye Disease

**To enroll in the program:**

- Complete the HIPAA form.
- If patients or their family members have questions, they can call (844) 405-1866 and enter the PIN# 200.
- Send a text message to the number (313) 284-4075. A Spanish-speaking representative will be available.

Once the patient completes and signs the form, we will send them instructions on how to use our services.

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

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

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

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

Once the patients and/or their family members submit the HIPAA form, they can scan and email any document for translations to Translator@BrombergTranslations.com. Examples of documents they can send for translation are:

- personal documents
- medical records
- forms and applications to government agencies and insurance companies

If you have patients that speak other languages, please reach out to Bromberg and we will assist you with your needs.
El idioma no debe ser una barrera para buscar atención médica, educación o servicios diarios importantes.

Con el objetivo de eliminar las barreras del idioma, Bromberg & Associates ha creado un programa gratuito, junto a Horizon Therapeutics, para ofrecer traducción e interpretación telefónica a personas y familias que hablan español y que tienen determinadas enfermedades.

Si usted o alguien de su familia tiene alguna de las siguientes enfermedades, puede recibir los servicios de traducción y de interpretación telefónica del equipo de Bromberg & Associates de forma gratuita:

- Enfermedades metabólicas poco comunes
- Enfermedades de los riñones poco comunes
- Gota no tratada
- Inmunodeficiencias primarias
- Enfermedad ocular de Graves y/o tiroidea

Para inscribirse en el programa:

- Complete el formulario digital de HIPAA
- Puede también:
  - Enviar un correo electrónico a Translator@brombergtranslations.com
  - Enviar un mensaje de texto al número (313) 284-4075. Un representante estará disponible para hablar con usted en español.

Una vez que complete y firme el formulario, le enviaremos las instrucciones sobre cómo usar nuestros servicios.

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

OPCIÓN 1: Hablar con un intérprete por teléfono.

Una vez que envíe el formulario de HIPAA, recibirá instrucciones para conectarse a una línea telefónica gratuita que le permitirá hablar con un intérprete profesional que puede ayudarle en consultas médicas, llamadas de emergencia, reuniones escolares, declaraciones y audiencias judiciales, y llamadas a organismos gubernamentales.

OPCIÓN 2: Si quiere traducir un documento.

Luego de enviar el formulario de HIPAA, envíe una foto de su documento por correo electrónico a Translator@BrombergTranslations.com. Algunos ejemplos de documentos que puede traducir son:

- documentos personales
- historias clínicas
- formularios y solicitudes a organismos gubernamentales y compañías de seguros
Miles’ Story
By Olivia Sirois

In September of 2021 we welcomed our second precious, healthy baby boy into the world. We were naive to the fact that our lives would forever change only 6 months later. In April 2022, Miles was diagnosed with Cystinosis. A disease that most people and even physicians have very little, or no information at all.

Even when Miles was an infant, he would spit up all the time, and never did I think anything of it - it seemed as though his older brother did the same. In the weeks approaching the diagnosis, my husband, and I noticed that he was falling behind in reaching his milestones. I remember telling my husband, “he just seems so sad”. He would lay his head on my shoulder as if he had no energy at all.

At Miles’ 6-month well-visit he weighed just over 12 pounds. He had fallen completely off the growth chart where he started in the 60th percentile at birth. The pediatrician told me to weigh him at home and if he continues to lose weight, call right away and they would see him and refer him to a GI specialist. By mid-April, Miles lost weight. I felt a sense of urgency to get him in to see the pediatrician ASAP. They said they would see him the next day. At the appointment he placed a GI referral and we saw them only a couple days later.

I remember walking into the children’s hospital not knowing what the outcome would be. I remember the physician coming in and telling me we would be staying in the hospital. He told me they would place an NG tube into Miles. I have a medical background, so I knew exactly what he was talking about. I’ve taken care of several patients who tolerate it and some who don’t. I just cried, knowing I would have to watch my child suffer, and knowing this was a more serious issue than what I originally thought.

We were in the hospital for two and a half weeks total. There were lots of tests being done and constant blood work being drawn to monitor Miles’ electrolytes, which were unstable.

About a week into our stay, the chief of nephrology came to see us. “We may have a diagnosis for your son. It is very rare, and we won’t know until it is confirmed through some genetic testing. It’s a genetic condition called Cystinosis”. Blood work was sent out and it was confirmed - our son has Cystinosis.

This rare diagnosis has changed our lives in ways we never could have anticipated. Being in the hospital, and especially the first several months at home, was emotionally overwhelming and physically exhausting. There were many tears shed and very little sleep.

We have adjusted our ‘new normal’ to several doctors’ appointments, weekly therapies, multiple medication administrations and feedings throughout the day.

It’s been a year and a half, and some days are harder than most. There is a sting of underlying grief that I believe will be present in my heart until I leave this earth. Watching my son through this process has given us much hope and he brings so much happiness to our lives and to the lives of those around him. We celebrate the victories that seem to come so easily for other children. I don’t think living with Cystinosis will ever be “easy”, for the patient or the family. But as we live our lives constantly adjusting to this new normal, the mountains don’t seem as overwhelming as they once were. Life is hard, but he is worth it.
The Cystinosis Research Network continues to utilize its financial resources to further its mission to secure a promising future for the cystinosis community through the support and funding of research grants that lead to improved treatments and ultimately a cure for cystinosis. Additionally, since 1996 CRN’s vision includes enhancing the quality of life for those with cystinosis. To that end, CRN expended significant resources in 2023 for the Nashville Family Conference which brought together hundreds in the cystinosis community along with both medical and industry partners.

Total income for the first seven months of 2023 was $461,964 which consisted mainly of grants received from our industry partners (a special thanks to President/VP of Development Jonathan Dicks) and $35,380 from fundraising. Expenses totaled $466,490 for a -$4,526 net income year-to-date. It should be noted that CRN’s expenses in a “conference year” are significantly higher than non-conference years and the majority of expenses were attributable to the conference and scholarships. Thanks to grants and fundraising by many in the cystinosis community, CRN’s current equity (assets minus liability as of July 31, 2023) stands at roughly $452,000 which is critical in funding additional research.

The CRN is a tax-exempt organization granted “501(c)(3)” nonprofit status by the I.R.S. The CRN Federal Tax ID # is 04-3323789.

I am so honored to be stepping into the role of VP of research. Christy has created wonderful partnerships with our fellow cystinosis advocacy groups around the world as well as an excellent working relationship with our pharmaceutical partners. I am excited to move forward in the coming years as more promising research continues to take place across the globe.

CRN has funded over $5.5 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, Germany 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. It’s our honor to collaborate with our international cystinosis advocacy colleagues to support the best researchers around the world.

**Current CRN Grant Commitments**

*Development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis*

*Drs. Katharina Hohenfellner and Julia Quitmann*

*Grant Amount: $155,075.09, two year study*

Patient-Reported Outcome measures (PROM) are questionnaire-based tools that can help healthcare professionals understand the health status or disease burden from the patient’s perspective. These tools can be used to evaluate new therapies or to improve the healthcare provided. Disease-specific instruments that measure health-related quality of life (HrQoL) is particularly informative, as they capture the needs and challenges of specific patient groups particularly well. As a multidimensional construct, HrQoL includes physical, emotional, mental, social, and behavioral components of well-being from the patient’s perspective. HrQoL can be measured using four different types of instruments: generic, chronic-generic, condition-specific and treatment-specific instruments. Generic questionnaires represent the full range of health conditions, address groups independent of their respective health state and are effective for comparisons between two cohorts (e.g., patients with cystinosis and healthy controls). Chronic-generic instruments are focusing on a chronic condition independent of its specific characteristics, while specific questionnaires are tailored to problems associated with a specific condition (e.g., cystinosis) or treatment (e.g. patients receiving a kidney transplantation). Despite the significant impairments experienced by patients with cystinosis, very few studies investigate HrQoL in this patient group and disease specific HrQoL measures are lacking. Thus, the primary aim of this planned study is to develop a PROM for children and adolescents with cystinosis. This instrument will capture the HrQoL from both the child/adolescent and parent perspectives. It will be applicable to clinical trials ranging from randomized clinical trials (RCTs) to surveillance designs, focusing on the impact of cystinosis and its treatment. The preparations have already started. We are currently developing the questionnaire “QUALIFY” (Health-related quality of life of children and adolescents with cystinosis) through intensive literature research and interviews with young German patients and their parents.

This preliminary version of QUALIFY needs to be cross-culturally validated in a larger sample (= investigated whether the instrument measures what it is supposed to measure) and adapted to the English, Spanish and French language. The study will take place in four phases within a 24-month timeline (Figure 1). Patients and parents will be recruited by both clinicians and patient organizations of the participating countries. In the first phase, the preliminary German version of QUALIFY will be translated into English, French and Spanish. In phase 2, these new versions of QUALIFY will be used in an online pilot test to analyze their preliminary psychometric properties. A cognitive debriefing will be conducted so that patients and parents can reflect on the comprehensibility, completeness, and applicability of the instrument. In the third phase, the refined questionnaires will be applied in an online field test to evaluate their final psychometric properties (incl. internal consistency, convergent validity, reliability). In the fourth phase, a final report and publications for scientific journals will be written. The final product will be a cross-cultural, psychometrically validated, practically feasible, and conceptually suitable instrument for children and adolescents with cystinosis. It will be available in English, Spanish and French for further (inter-)national studies. With further international collaborations, this tool can be linguistically validated and cross-culturally adapted for use in a wide range of countries. Although the project in this grant application refers to the development of PROM for children, the development of a PROM for adult patients is planned as a follow-up project.
Cognitive Control Systems in Cystinosis

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

Grant Amount: $315,193, two year study

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

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

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

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

Grant amount: $44,000, one year study

Nephropathic cystinosis lacks a practical therapeutic monitor for cysteamine therapy. The currently used white blood cells (WBC) cystine assay, although very specific for the disease, has many clinical and technical drawbacks limiting its routine use in most countries. Chitotriosidase enzyme activity in plasma is a potential alternative to WBC cystine in nephropathic cystinosis monitoring. Chitotriosidase is mainly produced in various tissues by activated macrophages upon stimulation by cystine crystals. We have previously demonstrated that the enzyme is significantly elevated in the plasma of cystinosis patients compared to both healthy and renal controls. 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-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. 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 Sanwal, 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.
Research Update, continued

of the disease and in particular of certain poorly understood cystinosis symptoms that appear to be caused by malfunctions other than the accumulation of cystine. In selecting this proposal for co-funding, the Boards of Cystinosis Ireland and CRN agreed that this is a scientifically significant proposal focused on a very important and strategic research topic for cystinosis patients. This project builds upon research and results generated from two previous projects co-funded by Cystinosis Ireland and the Irish Government’s health research funding agency (the HRB) – a project led by Professor Minnie Sarwal in UCSF, USA entitled “Targeting Autophagy in Nephropathic Cystinosis” and a project led by Professor Elena Levchenko in UZ Leuven, Belgium entitled “Unravelling the mechanisms of azoospermia and potential future treatments in male cystinosis patients”.

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

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

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

Total Grant: €10,000

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

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

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

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

This project proposes to generate isogenic immortalized epididymis and testicular cell models to study infertility associated with cystinosis, by using CRISPR/Cas9 technology to develop a deletion in the CTNS gene of normal human epididymis and testicular cells. The Sarwal group’s ongoing collaborations with Dr Smith at UCSF has enabled access to a rich tissue resource of normal testicular and epidydymal 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/Cystinosis Network Europe

I look forward to taking on the position 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/
Interested in taking part in a research study on fertility issues in males with nephropathic cystinosis?

About the Study: Dr. Minnie Sarwal (Nephrologist, UCSF), Dr. James Smith (Director of Male Reproductive Health, UCSF), and Dr. Polina Lishko (Human Reproductive Health Specialist, University of California Berkeley) are conducting a study to learn the cause of fertility issues in adult, male patients with nephropathic cystinosis who are treated with cysteamine. This study is currently jointly funded by Cystinosis Ireland (CI) and Cystinosis Research Network, USA (CRN). While the majority of men with cystinosis have not fathered children, there have been cases of some men becoming a father through assisted fertility. This has not been an easy journey and this study aims to find out more on the causes of male infertility in the hope it might help future treatment.

Primary Goals of the Study:
1. To better understand what causes male fertility issues in nephropathic cystinosis
2. To design novel therapies to potentially prevent young boys from losing male reproductive function, and
3. Patients with current infertility issues will be offered an option for sperm preservation for future use.
Follow-up to be discussed with Dr. Smith after initial visit.

What the Study Involves:
• Participation in this study would involve one visit to UCSF. The visit would include the following: Clinical examination, a blood test, ultrasound (of the testes which is a painless procedure similar to ultrasound used for pregnant women) examination for testicular volume and architecture, semen analysis, sperm motility (this means how fast sperm swim which can indicate the health of the sperm), morphology, the concentration of white blood cells, the level of fructose in the semen (semen is an organic bodily fluid, which contain sperms. Higher the absolute fructose concentration in the semen, lower is the number of sperms), and pH (pH is a measure of how acidic/basic water is. The pH of semen plays a crucial role in maintaining quality of sperm ensuring fertilization). Testicular and epididymal (coiled tube behind each testis) biopsies, sperm will be obtained by the TeSE (a procedure to collect sperm directly from testes), well-established techniques, performed routinely by Dr. James Smith. We will also collect semen via masturbation.
• All interested participants will have an online consultation via zoom with Dr. Smith to discuss this process prior to the visit to UCSF. Please see below the contact info if you would like to participate.
• If you are travelling to UCSF, part payment will be provided to help defray the cost of the travel. All clinic, blood draw, imaging and procedure costs at UCSF will be covered. For biopsies and TeSE local anesthesia will be used. When the biopsy is taken, you will feel pressure or minor discomfort. Mild narcotic to relieve the pain will be given. You would need to take it easy and rest for a week or two to recover fully.
• You will receive a free consultation with Dr. Smith, counselling for your results, and the test results will be available to your care team.

Benefits of Participating: Participation in this study will help us to understand the causes of male infertility in cystinosis. Understanding the cause will provide novel insights into new surgical and therapeutic approaches to either prevent or reverse male infertility in cystinosis. You will get a detailed report of your clinical, biochemical, and semen examinations. You will not be involved in treatment at this phase, however we hope that the results from this research will lead to the development of future treatment trials. All male participants with cystinosis will be offered the option of sperm preservation.

Who May Participate: You are eligible to participate if you are greater than 18 years of age (no upper limit) with a diagnosis of nephropathic cystinosis.

If you would like to participate in this study or if you would like more information, please contact minnie.sarwal@ucsf.edu or the study coordinator, Jim Cimino at 415/514.0192 or email to jim.cimino@ucsf.edu
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Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cystine, to accumulate in various organs of the body, including the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine. The availability of cysteamine medical therapy has dramatically improved the natural history of cystinosis so that well treated cystinosis patients can live into adulthood.

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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.