

THE CYSTINOSIS Advocate

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SAVE THE DATE!

2027 CRN Family Conference

Charlotte Marriott City Center
Charlotte, North Carolina
July 22-24, 2027

More details to follow soon at cystinosis.org

Rare Disease Week 2026



#RareDC2026 Recap

700+ Rare Disease Advocates **164** Patient Organizations

357 Meetings with Members of Congress

CRN's rare disease advocates gather in Washington D.C. to address their congressional representatives about the needs of the rare disease community during Rare Disease Week 2026 (see highlights on [Page 7](#)).

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THE PRESIDENT'S LETTER

Resilience in Action: Advocacy, Gratitude, and the Power of Community

Dear Cystinosis Community,

As we move into the spring season, I find myself reflecting on a year that has been both deeply personal for my family and profoundly meaningful for our broader cystinosis community. The journey that began last summer continues to shape how I think about advocacy, connection, and the responsibility we all share to keep pushing forward.

Many of you know that last summer marked a turning point for our family. In June, our daughter Elle began dialysis — a milestone we had long known was part of the cystinosis journey, yet one that is impossible to fully prepare for when it arrives. Just a few weeks later, in July, Elle received a life-saving kidney transplant from a deceased donor. The experience brought our family face-to-face with both the fragility of life and the incredible generosity that exists in the world.

Words will never fully capture our gratitude for the donor and family who made that gift possible. Their decision in a moment of unimaginable loss gave Elle a future, and our family a renewed sense of hope. Today, Elle continues to recover and grow stronger each day, and we remain deeply thankful for the care teams, friends, and this community who lifted us up during that time.

Our family's journey did not end there. In September, I had the privilege of donating a kidney to another child. While the paired exchange path we originally anticipated changed when



Elle and Finn Dicks enjoying life.

Elle received her transplant from a deceased donor, the decision to move forward with donation remained deeply important to our family. Through that donation, we were also able to participate in a voucher program that allows the gift of living donation to extend into the future. Because Elle went undiagnosed for nearly three years, the damage to her kidneys had well-advanced by the time we had answers, and she had complete kidney failure at just ten years old. That reality means she will almost certainly need another transplant later in life. The voucher now held in our family ensures that when that day comes, she has a priority pathway to a future living donor kidney.

Knowing that our donation could both help another child receive a life-saving transplant today and help safeguard Elle's future made the

The President's Letter, continued

decision profoundly meaningful. It was a powerful reminder that acts of generosity can create layers of impact — offering hope to another family now while also protecting the road ahead for our own.

That sense of shared purpose has also been front and center in our advocacy efforts this year. Members of the CRN community recently traveled to Washington, D.C. for Rare Disease Week on Capitol Hill, where patients, families, and advocates came together to share our stories directly with lawmakers. Standing alongside so many rare disease advocates reinforced an important truth: while each condition may be rare, our collective voice is powerful.

In addition, I had the opportunity to participate in both the FDA's Rare Disease Day and the NIH Rare Disease Day events in person. These gatherings bring together researchers, clinicians, regulators, and patient advocates to focus on accelerating research and improving access to treatments. Being in those rooms — representing the cystinosis community — serves as a powerful reminder that progress happens when science, policy, and patient voices move forward together. (For more Rare Disease Week updates see [Page 7](#)).

Moments like these underscore the mission of the Cystinosis Research Network: to ensure that families are supported, that research continues to advance, and that the voices of our community are heard wherever decisions about rare diseases are being made.

Our community has always been defined by resilience — parents



Jonathan Dicks at Rare Disease Week 2026 in Washington, D.C. advocating for the cystinosis community.

advocating for their children, adults with cystinosis leading the way for the next generation, siblings supporting one another, and researchers dedicating their careers to finding better treatments and ultimately a cure. This past year has reminded me that resilience isn't just about enduring difficult moments; it's about transforming those moments into action that benefits others.

As we look ahead, CRN remains committed to expanding advocacy, supporting groundbreaking research,

and strengthening the connections that make this community so extraordinary. Every family, every patient, every researcher, and every advocate plays a role in that progress.

On behalf of the CRN Board and my family, thank you for the support, compassion, and determination that continue to define this community. Together, we will keep moving forward.

With gratitude,

Jonathan Dicks

President | VP Development,
Cystinosis Research Network

Welcome New CRN Board Members

CRN is honored to welcome Frank and Hannah to the Board. Their leadership, dedication, and perspectives will help guide the organization as it continues to support families, advance research, and amplify the voices of those living with cystinosis. We are grateful for their willingness to serve and look forward to the impact they will make.

Frank Re, Treasurer

Frank Re is an LPL Financial Advisor with LCNB National Bank. He joined LCNB in July 2016 as a licensed administrative assistant and advanced to a fully licensed advisor in March 2019, a role he has served in since.

Frank is a graduate of the University of Cincinnati's Lindner College of Business, where he earned his Bachelor of Business Administration in Finance in 2016.

Frank was introduced to the Cystinosis Research Network through his local community and developed a deep personal connection to the mission after learning the story of a young girl living with cystinosis and witnessing her strength and resilience. Inspired by her courage, he became committed to supporting families affected by the condition and contributing wherever his skills and service can be of value.

He is grateful for the opportunity to serve as Treasurer of the Cystinosis Research Network and remains dedicated to advancing CRN's mission with integrity, accountability, and compassion.



Hannah Creel, Director

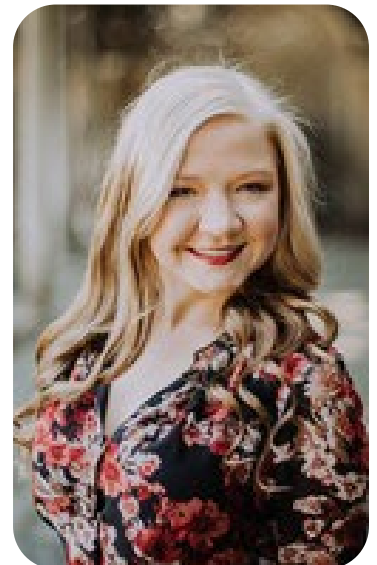
Hannah K. Creel is a licensed music educator and music therapy student committed to making music accessible, meaningful, and empowering for individuals of all abilities. She earned her Bachelor's degree in Music Education from Samford University with a vocal and choral concentration, and is currently pursuing board certification in music therapy through the University of Alabama's Equivalency Program.

Hannah has experience working with diverse populations including individuals with intellectual and developmental disabilities, autism spectrum disorder, dementia, and those in hospice and palliative care. She also teaches music at a school specializing in education for children with learning differences such as dyslexia, dysgraphia, dyscalculia, and ADHD.

Her clinical and educational work is grounded in a belief that music is for everyone and can be a lifelong tool for self-expression, emotional regulation, and social connection.

Her current education and research explores the role of music therapy in supporting executive function and emotional well-being in individuals with chronic health conditions. She is passionate about bridging clinical practice with research and music education to advocate for inclusive, person-centered care through Music.

Hannah lives with cystinosis and is excited to bring her personal lived experience and her professional skills to support the Cystinosis Research Network as its newest Director.



RESOURCES FOR

Patients and Caregivers

National Organization for Rare Disorders (NORD®)



NORD HELPS YOU NAVIGATE YOUR RARE DISEASE JOURNEY

Everyone who has a rare disease deserves the best possible care and support.

The National Organization for Rare Disorders (NORD®) is the first and leading U.S. organization to fight for improved quality of life and access to care for all rare diseases.

From day one, patients and caregivers have been NORD's top priorities, making us uniquely suited to serve your specific and complex needs. We provide financial assistance, caregiver relief, educational resources, webinars, and events.



Rare disease patients and families build a network of supporters by connecting at events like the Living Rare, Living Stronger® Patient and Family Forum.

"NORD's compassion and financial support has graciously afforded me insurance premium assistance and the ability to afford life-altering medications. NORD has given me the chance to live a normal life, and that gift of support changed my world!" - C.R., rare disease patient

Find Resources

For general and disease-specific assistance programs, search here:
rarediseases.org/patient-assistance-programs



Alone we are rare. Together we are strong.®

OUR IMPACT

(2018-2022)



\$174 million

in financial assistance for patients and caregivers



The NORD Helpline responded to nearly

700,000

requests for support



Nearly

2 million

Travel miles reimbursed for patients seeking treatment, clinical trial participation or specialist consultations



\$2.5 million

in non-medical emergency relief (ERF) provided to patients, including natural disaster and COVID-19 response funds since 2015



NORD®
National Organization
for Rare Disorders

Patient and Caregiver Assistance

We put patients and caregivers first. Our RareCare® Patient Assistance Programs – the first of their kind in the U.S. – are dedicated to supporting you and your caregiver team by providing:



Life-saving or life-sustaining medication, medical foods and formulas, and medical equipment



Financial assistance with insurance premiums and co-pays



Diagnostic testing assistance



Non-medical emergency assistance



Rare Caregiver Respite program (the first of its kind in the U.S.)



Travel assistance for clinical trials or consultation with disease specialists

At 8 months old, Lucia Karloutsos (bottom center) was diagnosed with Cardiofaciocutaneous syndrome. The family has found a partner in NORD to help them navigate their journey and is involved in advocacy through the Rare Action Network®.



Beyond financial support, NORD is the one-stop resource for information you need throughout your rare disease journey, such as:



The **NORD Rare Disease Database** is home to 1,300+ reports on rare diseases, with information on symptoms, diagnosis, treatments and resources to share with your health care provider. New reports are added each month in English and Spanish.



A free **online library of videos and classes** to learn about managing care, mental health, advocacy, participating in rare disease research, and more.



Conferences like the **Living Rare, Living Stronger® Patient and Family Forum**, where you can connect with others whose lives are also impacted by rare diseases (financial support may be available to attend; contact education@rarediseases.org to learn more).



Access to the world's top rare disease medical teams and clinical trials through the **NORD® Rare Disease Centers of Excellence Network**. Learn more: rarediseases.org/rare-disease-centers-of-excellence.

NORD's Helpline is here for you.

Reach us at **1-800-999-6673** or visit rarediseases.org/contact.

Helpline hours:

Monday–Thursday 8:30am–7:00pm ET
Friday 8:30am–6:00pm ET

ABOUT NORD

Since 1983, NORD has fought to improve the health and well-being of people with rare diseases by driving advances in care, research and policy. We're an independent and bipartisan nonprofit dedicated to reimagining a future where every person with a rare disease and their families live their best lives.



rarediseases.org

Alone we are rare. Together we are strong.®

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Persistence

By JB Shepperd



JB standing far right with rare disease advocates at Sen. Ted Cruz's office.

In the summer of 2025, I made the decision to attend CRN's Annual Conference in Grand Rapids, Michigan. The experience was life changing and gave me a sense of direction during a time where I felt that I had none. I was invited to speak on a panel of adults living with cystinosis, and I loved every minute of it. It was incredibly fulfilling having the opportunity to speak on my experiences and provide a sense of comfort to current patients and parents of younger patients. Never in my life had I felt something like that. So, when I was asked if I was interested in speaking on Capitol Hill in favor of rare disease legislation, I agreed with no hesitation. I was hungry to exercise my public speaking

skills and honored to have the chance to help make real, concrete change in this country.

The event was everything I expected and more. I had an incredible time with Ella and Jeff, the other family attending the event, and learned so much about myself; what it means to be an advocate, and how to navigate the advocacy process. Much of the first few days there were focused on preparation and networking, both of which I was very excited to get into from the beginning. I met so many wonderful people with inspiring stories that I will never forget, just by sitting at random tables during the breaks between speaking events. The energy in the room was palpable; there were so many amazingly strong individuals

all in one place, all ready to share their stories.

I was a bit more nervous for the congressional office meetings themselves, although after the days of preparation, I felt more than capable of expressing legislative asks, as well as their connection to my own story. And every meeting went well! I found every staffer we met with to be receptive to our talking points and easy to converse with. I had a wonderful time getting to know my fellow advocates coming from my home state and had a blast collaborating to make a plan for how we would present ourselves and our asks. For me personally, I feel that this whole event has set me on a path towards advocacy and a life in

Persistence, *continued*



JB, (fourth from the left), and fellow Texas advocates meet with Rep. Chip Roy's office staff.

“ I feel that this whole event has set me on a path towards advocacy and a life in public speaking that would have been difficult to find otherwise. ”

public speaking that would have been difficult to find otherwise.

For anyone who may be reading this and wondering if advocacy is right for you, I strongly encourage you to give it a shot. Even if you are not the most practiced speaker or consider yourself to be a bit shy, the important thing to remember is that in situations like these, our strength comes from persistence. I fully recognize that my

recent efforts at Rare Disease Week may not yield direct change in the next year. Or the next 5 years, or maybe even 10. But change is built upon the shoulders of those who came before you, and those who will come after you. Even if you don't want to speak, simply your presence at events such as these, supporting those who do decide to speak, is building upon the foundation that was there before you arrived. Together, we are creating a

bonfire that cannot be ignored, but without those who add fuel to the fire and those who fan the flames, we will not be noticed. So please, do your part, add your branch and keep the flames alive for those who did the same for you and those who will follow in your footsteps. Thank you to everyone at CRN for this incredible opportunity. I hope to be adding my fuel to the fire for the rest of my life.

Speaking Up for Rare: Our Journey to Rare Disease Week in Washington, D.C.

By Jeff Zich, with reflections from Ella Hensel

My name is Jeff and I have a 14-year-old daughter, Ella, who was diagnosed with cystinosis at 18 months. We had the pleasure of attending our first cystinosis conference last July in Grand Rapids this past summer. What a great event where Ella was able

to shine. She shared her experience with cystinosis in front of the whole conference as part of the Speak Up, Speak Out Program. After expressing our interest to get more involved, we were invited to attend Rare Disease Week in Washington, D.C. I want to

thank Clair Johnstone and Jonathan Dicks for the opportunity to attend such an important and wonderful event.

At times, the anticipation leading up to Rare Disease Week was a bit stressful for me, but not for Ella. She

Speaking Up for Rare, *continued*

was a champ: calm, put together, and nonchalant. Preparation involved many moving pieces: Zoom calls, planning the week, and keeping a close eye on our inbox for updates.

We chose to leave on Sunday to begin the drive from Illinois to Capitol Hill. Due to a snow storm the 12-hour drive turned into a 17-hour white knuckled journey, but it was worth it!

We arrived in D.C. on Monday and rested before meeting up with Jonathan, President of the CRN. He's a dedicated man when it comes to advocating for cystinosis. He also has a daughter with cystinosis. I first met him at the July conference, and both Ella and I knew he was a good-hearted soul. At dinner, we got to know more about his life and his family's journey. When the dinner ended, any stress and nervousness melted away. That dinner had a way to make even a high-tension guy like me relax.

Tuesday started with an Uber ride with Jonathan, and meeting up with JB who was joining us for the week. JB is 25 years old and lives with cystinosis. We arrived at the Ronald Reagan Building, welcomed by a huge reception hall surrounded by flags from around the world. Sessions started with discussions on the importance of being there to advocate for our rare disease community with our Representatives and Senators. We learned that in our state of Illinois, there are over one million people with a rare condition. Even though the cystinosis community is a fraction of this, our voices needed to be heard. We had the pleasure to watch videos



Ella and her dad, Jeff attending Rare Disease Week 2026 in Washington, D.C.

***30 million
Americans live with
a rare disease,
representing just
under 10% of the
U.S. population.***

made by some of the "Rare Art" creators. Rare Art is a campaign that showcases and honors rare disease artists. Ella and I were fortunate enough to meet a few of them, like Emily, a 15-year-old from Alaska who lives with a dermatological disease that affects her skin and teeth, and Wes, the creator behind characters such as Po from Kung Fu Panda and Oscar, the lead in Shark Tale, whose rare disease eventually forced him to retire from DreamWorks. We also met so many others who are passionately advocating for the rare disease community.



Jonathan Dicks, president of CRN, Ella Hensel, with her dad Jeff Zich, and JB Shepperd recently advocated for the cystinosis community at Rare Disease Week 2026 in Washington, D.C.

Tuesday evening's reception was incredible! We had the chance to meet so many different people from across the country. Ella kept her cool, even though I could tell she was exhausted. Once we got to the hotel that evening, we were zoned out...time for some sleep.

Wednesday was long and demanding, but highly educational. At the Ronald Reagan Building, we heard rare disease advocates emphasize the critical role of genetic and genomic testing, clinical trials, and funding for new therapies.

***There are just over
700 treatments
available for the
10,000+ rare disease
worldwide.***

Speaking Up for Rare, *continued*



Ella and Jeff with fellow advocates and Representative Brad Schneider's aide.

The Meeting hosts provided us with snack areas and a nap nook. With Ella likely needing a kidney this year, she is at the tired all the time stage. She took full advantage of the nook. While she rested, I got to watch a video of a trial drug that literally turned around an entire rare disease community, to give those young men a better quality of life. This series of meetings reinforced the power of advocacy on The Hill. As a rare disease community, we are strong and represent a significant constituency in our lawmakers' districts.

We were then organized into groups by our home states. We worked with new friends to decide who would speak first, how long, and what our 'asks' were. Once we were in front of each Representative. Ella and I both asked for May 7th to be officially recognized as Cystinosis Awareness Day. I lobbied for the Credit for Caring Act (H.R. 2036/S. 925) to help caregivers retain their jobs, if they

are caring for a loved one, along with financial help and related support. As a caregiver to Ella when she was diagnosed, this hit close to home.

As Wednesday came to a close; we sat down for dinner with JB. We talked about what to expect for Ella's transplant, and how she would be affected with a new kidney (JB has already received a transplant). JB was open and honest in sharing his experiences. Ella and I are so grateful to have met this young man. We found out he'd recently moved to Chicago, less than an hour from us!

...to rise up about our disease and to not hide in the dark, that's what brought me here.

- Ella

That brings us to Thursday...THE DAY. This was the whole reason we were in our nation's capitol for this fantastic event: to speak directly with our legislators. Our day began with a meeting with Rep. Mary Miller's staff. They listened, took notes, had an open heart and were kind to Ella and the other young lady who was advocating. Next, we headed to another building to meet with Raja Krishnamoorthi's office. The walking was already starting to take its toll on Ella, but she is such a trooper. Once there, we had the chance to talk to a member of Rep. Krishnamoorthi's staff. We were slightly disappointed we didn't get to speak directly to any of our lawmakers. Because of the timing of elections and those campaigning, it was explained to us that only a few Representatives and Senators were currently in D.C.

During lunch in the House building, we saw a large number of uniformed Air Force members, which was an impressive sight, especially for Ella.



Ella making new friends in Washington, D.C.

Speaking Up for Rare, *continued*



The rare disease constituency of Illinois representatives.

“ *It was such an honor to attend Rare Disease Week and to meet lots of people who are just like me...willing to share their story.* ”

- Ella

At the Senate building we were met by all of the Illinois rare disease advocates and were able to talk to senior staff here. We asked the Senior Advisors in Senator Tammy Duckworth's and Senator Dick Durbin's offices about supporting Cystinosis Awareness Day. The gentleman of Senator Durbin's Health staff was attentive and very responsive. Ella was so cool as if she'd done this before; I am so proud of her.

We left the Senate building for one last meeting, and Ella said goodbye to a couple of young people she met.

Once through security, we were able to reach Rep. Brad Schneider's office. His senior staff member heard us out and let us take a photo with her - the only staff that allowed it throughout the day.

Up next was a scheduled group photo which was eventually canceled due to the rain. While waiting, we met a brother and sister with different rare diseases and took a few personal photos once the rain stopped. At the closing banquet, Ella, though exhausted, said goodbye to her new acquaintances on the way out. After dinner with our cystinosis friends we

headed back to the hotel and pretty much passed out cold.

The following day, we went our separate ways with Jonathan heading to Rare Disease Day at the NIH, and JB flying home. We offered JB a ride to the airport and found out that the National Mall isn't a shopping center, but a large stretch of land filled with monuments and memorials. Ella and I bid farewell to our new friend, JB, and set off.

On our way home we agreed to visit the D.C. Zoo, where the star of our day was the pandas. Ella's delight was contagious. Seeing them play directly in front of us, bouncing around on the branches of his tree had Ella beaming!

I have to say, once again, how grateful we are to have been invited, and able to attend this year's Rare Disease Week. It was an absolute honor and privilege. Jonathan was the host of all hosts: kind, knowledgeable, and kept us on schedule. I've never had an experience like this, and I know Ella hasn't either. It was amazing. It was informative. We want to thank the CRN for the pleasure of going to Rare Disease Week 2026.

“ *Thank you for this life-changing opportunity, CRN!!!* ”

- Ella



It's Back!

July 17 - July 19, Chicago, IL



Watch videos from the 2025 Speak Up, Speak Out: Cystinosis and Me on the Cystinosis United Instagram Channel.

LED BY **Believe**LIMITED WITH



CYSTINOSIS RESEARCH NETWORK

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2026 CRN Academic Scholarship Application Information

By Gail Potts, Director

The Cystinosis Research Network has been offering academic scholarships for 20 years. The Deanna Lynn Potts Scholarship was first awarded in 2000. These awards are for individuals with cystinosis, as well as one for a sibling of an individual with cystinosis. We are proud to be able to ensure our warriors have a little easier journey as they pursue their careers. It has been a thrill to see these young people achieve their dreams and succeed in their chosen fields. The scholarship awards are for \$2,000.

Applications for the 2026 Academic Scholarships will soon be available on our website. The application process is rather simple. You will be asked which scholarship you are applying for: the Individual Living with Cystinosis, the Deanna Lynn Potts Scholarship for an individual with

Cystinosis, or the Sierra Woodward Sibling Scholarship for a sibling of an individual with Cystinosis. We are asking you for identifying information about you, and where you intend to go for your education, college or vocational school. You will be required to include an essay answering two questions. Once the committee receives the application, we will contact you to let you know we received it, and request a letter from your doctor confirming your cystinosis diagnosis, a transcript from your high school or college, and two letters of recommendation from teachers, guidance counselors, and or clergy. The deadline for all applications is August 1st. The committee will convene and make their decision on the recipients of the awards.

The essay will be judged on the

basis of rationale, grammar and comprehension. Transcripts and letters of recommendation will be considered in the final decision with Grade Point Average (GPA), courses taken and class standing used as part of the evaluation. The CRN Board will establish an independent judging panel to evaluate and rate the applicants. Finalists may be interviewed before selections are made. The decisions of the judges are final.

Once you have received an award we will consider a second award after two years. At which time, you can apply again.

For information and applications, check our website at: cystinosis.org/scholarships or contact Gail Potts at gpotts47@yahoo.com.



Program Status:
OPEN APPLY NOW at
www.mygooddays.org/apply

Assistance Amount:
\$ 7,500 per year

Good Days is a national non-profit charitable organization that lifts the burdens of chronic illness through assistance, advocacy, and awareness.

We make life-saving and life-extending treatments affordable and act as your advocate, helping you navigate the system and guiding you to additional support through foundations and other organizations dedicated to those with specific, life-altering conditions.

Learn about our copay, travel and premium assistance programs to support individuals with Nephropathic Cystinosis.

Contact us for eligibility details. Ask a Good Days Patient Care Navigator for more information toll free at (855) 327-1134 or at <https://tinyurl.com/GoodDaysCyst>

CAMP CONNECTIONS

let's go on a virtual adventure!

American Kidney Fund®



Camp Connections: Cystinosis,

offers a special quarterly virtual gathering for kids living with this rare disease, bringing campers from across the country together through Zoom to build friendships with peers who truly understand their journey. With interactive activities like arts and crafts, STEM projects and game nights, campers can enjoy moments of joy, creativity and connection from wherever they are.

These quarterly sessions are part of **Camp Connections**, an innovative monthly virtual program hosted by the American Kidney Fund (AKF) that provides pediatric kidney patients with opportunities to connect, create and thrive alongside peers who share their experiences. Every new participant receives a welcome swag bag, along with monthly activity kits to kick off their camp adventure.

TO LEARN MORE ABOUT CAMP CONNECTIONS, PLEASE VISIT
WWW.KIDNEYFUND.ORG/CAMP-CONNECTIONS
OR REACH OUT WITH QUESTIONS!
CAMPCONNECTIONS@KIDNEYFUND.ORG

Muscle Weakness and Cystinosis, My Story

By Paula Shal

Editor's note: *It is a privilege to share Paula's remarkable cystinosis journey. Her resilience, courage and willingness to share her story is heartwarming. It should be noted, however, that she was not diagnosed until age 17, and did not receive cysteamine therapy until then. This impacted the severity of her symptoms and may not represent the typical disease progression in children diagnosed in infancy and treated with cysteamine from the time of diagnosis.*

Hi, my name is Paula Shal. I am 55 years old, and I have cystinosis. I was diagnosed with cystinosis when I was 17 years old. I have infantile cystinosis, but my crystals did not accumulate aggressively. The only symptoms I had were photophobia that presented itself when I was about five years old. I was also short in stature, but other children my age were the same size. I was a picky eater but nothing crazy. My weight was proportionate to my height. I

never faulted my parents for not taking me to an eye doctor when my photophobia began. Who would suspect that a child who was sensitive to light would have a rare disorder that damages organs and muscles?

My journey with cystinosis has been long and complicated, but I want to focus on muscle weakness and how it has impacted me. As you read my story, please keep in mind that I did not have any cysteamine therapy for 17 years. But also keep in mind that my cysteine crystal accumulation has not been as aggressive as some. I don't quite know how to interpret that, but I know that having cysteamine therapy would have prevented many of my issues. From the perspective of a 55-year-old with multiple organ failure and neuromuscular weakness from cystinosis, I can't stress enough the importance of being compliant about taking Cystagon or Procysbi and getting cysteine levels checked periodically so that dosage can be

adjusted.

Throughout my school life, I was never as strong as my peers. Gym class was hard (unless we were square dancing or using the parachute). I played little league softball in grade school, but I wasn't good. When the other kids far surpassed my physical abilities, I didn't play any sports (except for one disastrous stint on the eight-grade volleyball team). I tried out for cheerleading, but was told my voice wasn't loud enough. I took piano lessons, but it became increasingly difficult to reach the keys. I sang in the chorus and participated in school musicals, but my voice was starting to crack, and I couldn't hit the notes I once could.

I was pushing through life, trying to participate in everything, not knowing I had a rare disorder that caused neuromuscular weakness. I couldn't tell people, "I have this disorder, and this is why I am the way I am." Side note: I also knew my kidneys were failing but didn't know why. At times I felt frustrated, but my motto has always been, "If you are unable to do something, find something you CAN do." I excelled in academics, drama, student leadership, and was editor of the school newspaper. I enjoyed supporting my high school's sports teams even though I couldn't participate. I was fulfilled.

When people think of muscle weakness from cystinosis, they may not realize the impact. I was followed at the NIH for many years and was told the muscle weakness tends to start in the hands and work its way to the respiratory system. I would say this is true in my case. The muscles in my upper back, feet,



I had a pretty healthy childhood.

Muscle Weakness and Cystinosis, My Story, *continued*

neck, face, vocal folds, esophagus, and GI system are also significantly affected. I was also told cysteamine therapy would halt or slow down the muscle wasting. I believe this was also true in my case, but remember, I didn't take cysteamine for 17 years. I confess there were also short periods of time when I didn't take it for various reasons. When I met my husband, I would skip doses, so I didn't smell. If I am feeling nauseous for other reasons, I don't take medicine that also can make me nauseous. Because of muscle weakness, I have aspirated when I vomit and ended up with pneumonia. This can be extremely dangerous with a compromised respiratory system, so I have to weigh the risk.

My muscle issues were status quo for many years. I took a fitness class and ice-skating class in college and backpacked through Europe. I have hiked Diamond Head, Sedona, The Virgin Islands National Park, and so much more. I am never the fastest, but I finish. I get into good workout routines, but when life gets overwhelming, they are usually the first to go. Despite my best efforts, my neuromuscular weakness from cystinosis has slowly progressed. I guess in some ways I became "used" to managing my deficits. I eat and drink slowly, so I don't choke. Our house is set up to adapt to my abilities as much as possible. I try to find kitchen, office, and beauty tools that are easy to use. I hoped I would remain stable. But my muscle weakness has escalated in the past ten years. Even though I knew I would probably get weaker, I feel a little blind-sided by some of the challenges I have had, which is why I want to share them.



With my husband, Jim (and Snickers) enjoying Florida in 2026. Travel can pose some challenges. I have PTSD from landing in unfamiliar emergency rooms while travelling. But I've learned my limits, and we make it happen to maintain our mental health.

I worked for 23 years in a corporate job where I spent much of the day on a computer. I had several side hustles and did volunteer work from home, which also required time on a computer. One night after a long work week, I pinched a muscle in my upper back. I have terrible posture from muscle weakness, and this can cause pinched nerves. I didn't know what I didn't know. I was in agonizing pain that shot down my right arm. I am right-handed and couldn't use my arm or even type, which was essential for my job. I went to physical therapy and soon regained function...until I pinched it again. The second time I pinched it was on the same day my dad passed away unexpectedly. I made the decision to go to the emergency room to get treatment, so that I could get through the week. They did a CT scan and prescribed steroids and painkillers. And I went back to physical therapy.

I was so weak that I continued to

pinch the nerves two more times. The last time I couldn't recover fully, and I can now only lift my right arm to my chest. Continuing PT at home has helped me avoid additional damage, but losing this function along with only having about 25% function in my hands has had a big impact on my quality of life. I can't do things like putting a leash on my dog to take her for a walk, locking/unlocking certain doors (I'm terrified I will lock myself in a public bathroom), and opening just about anything that requires fine motor skills. It is discouraging because I like to be independent, and hate bugging my husband for help. And I want to walk my dog.

My most significant challenge in the past ten years has been muscle weakness in my respiratory system. I have always been short of breath. It is why I hated gym class. Running laps put me in a panic. I thought I would remain stable if I took my cysteamine therapy. But again, I didn't have

Muscle Weakness and Cystinosis, My Story, *continued*

“If you are unable to do something, find something you CAN do.”

cysteamine therapy for 17 years. Still, I felt a little blindsided because I was stable for so long.

There was a time when I was hospitalized for a UTI, and my oxygen saturation level dipped into the upper 70s in the middle of the night. A nurse came running into my room to check on me. No one ever suggested I follow up. My husband was also noticing that I would stop breathing in the night. I knew that other people with cystinosis used CPAP and BIPAP machines and that sleep apnea could be an issue. I asked my primary physician for a referral for a sleep study. I was a 100-pound woman, and she said I likely didn't have it but reluctantly gave me a referral. Long story short, I now see a Pulmonologist who specializes in sleep medicine and neuromuscular weakness.

I am not a doctor, and don't claim to know everything, but I feel this is really important information that I have found to be true:

- Most pulmonologists don't understand neuromuscular weakness in the respiratory system. Their expertise is geared toward the lungs. In my case, my lungs are healthy. It is the muscles used for breathing that are weak—my diaphragm, intercostal muscles (muscles in the ribs), and neck. It is important to find a pulmonologist who understands this.

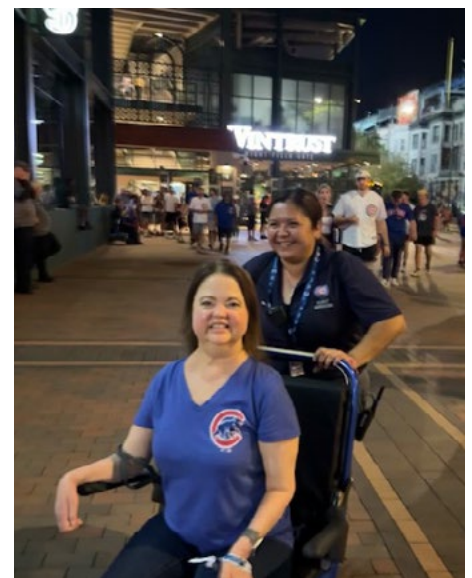
- CPAP is not an appropriate treatment for someone with respiratory weakness. CPAP stands for Continuous Positive Airway Pressure. The key word is “continuous.” If air is continuously pushed into the lungs of someone with respiratory weakness, they can't exhale fast enough, resulting in low oxygen levels, which can be dangerous. BIPAP is a more desirable treatment of mechanical ventilation because settings can be adjusted so that the air is not continuous.
- Neuromuscular weakness is not the same as obstructive sleep apnea, but both can occur at the same time.

When I recline or lie down, I feel like an elephant is standing on my chest. I become short of breath quickly, and my heart rate skyrockets. If I stayed in this position, I would go into respiratory arrest. I receive ventilation from a BIPAP, which takes over my breathing so that I can recline and lie down. I use a mask with nasal pillows as opposed to a full-face mask. It was an emotional adjustment starting the BIPAP. I got used to it, and I love being able to breathe better, but just a few years ago I had some breathing episodes that left me hospitalized multiple times. My breathing just shut down when I was in the sun, heat, and/or humidity. I couldn't move, and my chest was tight. At first, I thought I was dehydrated, since that is a common occurrence for me and many with cystinosis. But it seemed different.

I had a battery of tests, and doctors were perplexed. My pulmonologist's solution was, “use your BIPAP more often.” I didn't like this solution. I kept pushing forward thinking that

activity and exercise would help, but I just kept getting worse. Each time I went outside that summer, I couldn't breathe—even if I just opened the door to let my dog out. It escalated so much that I couldn't breathe inside either. My days were filled with gasping for air and high heart rates. My body felt heavy. I remember feeling accomplished when I could get up to put something in the garbage can. I had to wear my BIPAP for almost 16 hours a day. I started having panic attacks, especially at events like concerts and baseball games. I had to be hauled away in a wheelchair and/or use my BIPAP to recover. The wheelchair caused motion sickness and more panic attacks because I was afraid I would vomit and aspirate. All this time my oxygen saturation level was normal at between 95 and 100.

My mom was in long term care, and I didn't want her to see me living ventilated. My husband would push



I was wheeled out of Wrigley Field in 2023 after a challenging night of trying to breathe. We learned to take my BIPAP with me wherever I go, and we now take our own wheelchair. I'm smiling because the Cubs won.

Muscle Weakness and Cystinosis, My Story, *continued*

me in a wheelchair with the BIPAP on to get to her door. This saved my energy. Then we would leave the BIPAP in the wheelchair outside the door. I could go into the hallway and take a few puffs of air when I needed it. I thought this would be my new normal. I was trying to figure out how to eat and wear a BIPAP. I finally tried something new: Just stop and rest, become a couch potato. My theory was that I may have injured a breathing muscle, and it needed to heal (???) I did get better and started moving more again, but I have never been back to where I was. I've just learned my limits such as not to push my activity too far, or spend more than a couple minutes in hot sun or humidity.

I know so many people, with and without cystinosis, who are reluctant to get a sleep study. If you or someone you know with cystinosis has any muscle weakness at all, I encourage a sleep study. I would also ask for Pulmonary Function Tests (PFTs) and get them periodically. My PFTs show that I am breathing at 44% of what I should be. I have learned that when a PFT goes under 50%, that is usually a sign that mechanical ventilation is needed. Breathing properly has many benefits such as better sleep, cognitive function, and cardiovascular function. I had the realization one day that neuromuscular weakness from cystinosis is similar to muscular dystrophy. I found some wonderful resources in the muscular dystrophy community that have given me a wealth of information. One of my favorites is <https://breathewithmd.org>. There is also a Breathe with MD Facebook group. The admin. and members really know their stuff and can provide doctor recommendations.

“ *I have met some of the most incredible people in this community. Our (cystinosis) community lifts each other up by being compassionate and empathetic, which has helped me to be more compassionate and empathetic in daily life.* ”

This resource from the American Thoracic Society also provides a great overview of respiratory neuromuscular weakness—<https://www.thoracic.org/patients/patient-resources/resources/neuromuscular-weakness-adult.pdf>.

When I was in my 30s, I asked for a referral to physical therapy. I wanted to be proactive and thought this would keep me motivated. The physical therapist assessed me and thought I was a little weak, but because I wasn't in pain or significantly disabled, she gave me exercises to do at home and sent me on my way. I wish I would have pushed this further. Physical therapists can target specific areas that need strengthening. I really needed to meet regularly with a physical therapist who kept me accountable. I may not have pinched nerves if I had utilized PT earlier. I have gone to speech therapy for my voice and swallowing, and occupational therapy for my hands and have found them all helpful. I have also found that the PT I do for my pinched nerves strengthens my neck and intercostal muscles, which has improved my swallowing and breathing very slightly.

I chose to focus on my muscle weakness issues because I feel like muscle wasting gets less attention in the cystinosis community than

issues like kidney transplants. For the record, I have received four kidney transplants from four living donors—my mom, brother, husband, and a friend. My brother and husband's kidneys were not a match for me at all. In today's world, a transplant team probably would have excluded them from being my donors. I had my brother's kidney for eleven years and my husband's kidney for seven. I have had my friend's kidney for the last 17 years, but it has been slowly declining for the past eight years and is now functioning at 15%. I've grappled with the decision of whether to live out the rest of my life on dialysis or try to receive another transplant (more on this later). Dialysis was not kind to me in the past, but surgery is risky primarily because of my breathing issues. I would have a breathing tube during the surgery, but as soon as it is pulled out, someone needs to immediately get my BIPAP on me. This is not something that a surgical team is accustomed to doing. Anesthesia can also suppress breathing further and cause nausea—my worst enemy!

One of my heroes in the cystinosis community, Jessica Jondle, once wrote a blog entry about how she was grateful to have cystinosis. Her elegant prose about her feelings have

Muscle Weakness and Cystinosis, My Story, continued

stuck with me. I reflect on that blog often and ask myself if I am grateful for having cystinosis. The answer is that I would rather not have it, but I appreciate the lessons this disorder has taught me. I have met some of the most incredible people in this community. Our community lifts each other up by being compassionate and empathetic, which has helped me to be more compassionate and empathetic in daily life. Parents of children and adults with cystinosis have taught me what my parents went through. My parents didn't talk to me about it, but now I have a better idea. I have observed children and adults with cystinosis exhibiting so much resilience, and it is a reminder for me to keep fighting.

I am also thankful to all the health care professionals, researchers, and advocates who work tirelessly to understand cystinosis and organ transplants and fight for better treatments. I am grateful for my kidney donors, and all living and deceased kidney donors. I am grateful for people who check in just to see how I'm doing. I am grateful for people who have treated me as an equal even though I am a quiet woman with physical differences who manages a chronic illness and disability. I am grateful for my BIPAP because the previous treatment was an iron lung. I am grateful to my loving parents who endured many doctor appointments and hospitalizations with me. They were only mildly overprotective, but for the most part, treated me like they did the rest of their children and encouraged me to go out into the world on my own (although I KNOW my mom was ecstatic when I married a good husband).

And my husband. I'm not sure where

I would be without him. He has been my care partner for over 25 years and knows my body almost as well as I do. He very patiently listens to me complain. He helps me problem-solve my known issues and helps me determine when I need medical care. Muscle weakness often makes it difficult to use the tools I need to take care of myself. He helps me by changing the water in my BIPAP, taking it apart and cleaning it, administering Cystadrops, changing my Dexcom monitor that monitors my blood sugars, injecting my Procrit to treat anemia, and so much more. I am grateful for the adventures we have had and wonderful memories we've made that would be difficult to replicate now.

After a long career, my job now is to stay alive. After an internal battle, I made the decision to get tested to see if I am a candidate for a fifth transplant. It feels selfish when some people can't get one transplant. But after all my husband has done for me, I need to be here for him, and as healthy as possible for as long as possible. I have been told I am a candidate, but I need one more test that can't be done until my kidney completely fails because the test utilizes dye that would damage my kidney. Kidney failure itself also affects breathing, and I dare to dream that my breathing might improve a little with a healthy kidney. The hope of a transplant keeps me going even though it is also a scary thought. I have to try.

I don't dwell on what my life would be like without cystinosis, but sometimes I mourn what might have been. My motto of "If you are unable to do something, find something you CAN do" is becoming increasingly difficult.

| Medicine | Food |
|--|---|
| <input checked="" type="checkbox"/> 3 Dexcom sensors | <input checked="" type="checkbox"/> Cookies/chocolate |
| <input checked="" type="checkbox"/> Insulin pens | <input checked="" type="checkbox"/> Nuts/Craisins |
| <input checked="" type="checkbox"/> Insulin needles | <input checked="" type="checkbox"/> Ritz |
| <input checked="" type="checkbox"/> Test strips | <input checked="" type="checkbox"/> Wheat Thins |
| <input checked="" type="checkbox"/> Lancet | <input checked="" type="checkbox"/> Egg salad sandwiches |
| <input checked="" type="checkbox"/> Pill boxes (filled) | <input checked="" type="checkbox"/> Pick up Poke |
| <input checked="" type="checkbox"/> Prescription bottles for boxes | <input checked="" type="checkbox"/> Tortilla shells |
| <input checked="" type="checkbox"/> Procsybi | <input checked="" type="checkbox"/> Tomato sauce |
| <input checked="" type="checkbox"/> Levothyroxine | <input checked="" type="checkbox"/> Shredded cheese |
| <input checked="" type="checkbox"/> Astral and Resmed machines | <input checked="" type="checkbox"/> Taco seasoning |
| <input checked="" type="checkbox"/> Distilled water | <input checked="" type="checkbox"/> Bread |
| <input checked="" type="checkbox"/> Cough Assist | <input checked="" type="checkbox"/> Peanut butter |
| <input checked="" type="checkbox"/> Zofran | <input checked="" type="checkbox"/> Blueberries/apples |
| <input checked="" type="checkbox"/> Dramamine | <input checked="" type="checkbox"/> Drinking water gallons |
| <input checked="" type="checkbox"/> Culturelle | <input type="checkbox"/> OJ |
| <input checked="" type="checkbox"/> Xylmelts | <input type="checkbox"/> Fiberone Cereal |
| <input checked="" type="checkbox"/> Imodium | <input checked="" type="checkbox"/> Oatmeal |
| <input checked="" type="checkbox"/> Claritin | <input checked="" type="checkbox"/> Granola bars |
| <input checked="" type="checkbox"/> Tylenol | <input checked="" type="checkbox"/> Dates/walnuts |
| <input checked="" type="checkbox"/> Gas-x | <input checked="" type="checkbox"/> Brown sugar |
| <input checked="" type="checkbox"/> Band-aids | <input type="checkbox"/> Butter |
| <input checked="" type="checkbox"/> Saline spray | <input checked="" type="checkbox"/> Coffee/filters |
| <input checked="" type="checkbox"/> Cefpodoxime | <input checked="" type="checkbox"/> K-cups |
| <input checked="" type="checkbox"/> Mucinex | <input checked="" type="checkbox"/> Aluminum foil |
| <input checked="" type="checkbox"/> Lorazepam | <input checked="" type="checkbox"/> Cheese |
| <input checked="" type="checkbox"/> Face prescriptions | <input checked="" type="checkbox"/> Gatorade |
| <input checked="" type="checkbox"/> Prescription shampoo | <input checked="" type="checkbox"/> x CARTONS |
| <input checked="" type="checkbox"/> Vicks | <input checked="" type="checkbox"/> x 1/2 bottle silverware |
| <input checked="" type="checkbox"/> Extra Face mask for bipap | |
| <input checked="" type="checkbox"/> Blood pressure cuff | |
| <input checked="" type="checkbox"/> Thermometer | |
| <input checked="" type="checkbox"/> Pulsox | |
| <input checked="" type="checkbox"/> eye drops | |

"Tell me you have cystinosis without telling me you have cystinosis." I keep a word document of things we need to remember to pack when we travel. My medical needs require a suitcase that is larger than my clothing suitcase.

I can't fly because I can't breathe at high altitudes. I could take my BIPAP, but if it malfunctions, then I am the reason the plane has to make an emergency landing. I'm not ready for that. I miss being able to lie down on the couch. I worry that if I fall and can't get up, I will stop breathing. I want to eat and drink without worrying about choking. I miss going for long walks and hikes. I wish I could sing, scream, play an instrument, learn sign language, climb multiple flights of stairs, button buttons, put on jewelry, paint my nails, spend a day in the sun, take care of other people, and independently walk my dog...

Muscle Weakness and Cystinosis, My Story, *continued*

but I am so appreciative, sometimes in awe, that I am still here. I get to see seasons change, flowers blooming, sunsets. I get to hear birds chirping and children laughing. I get to smell coffee in the morning and lilacs in the spring. I can walk. I can talk. I can see. I can eat. I often think of those with cystinosis who are no longer here. Some friends. Some acquaintances. All fighters. I grieve for them and keep their families in my prayers. I try not to take life for granted. But I'm human, and I don't always succeed. I like to think that

“The discipline required to manage cystinosis has benefited other areas of my life. I believe in the paradox that two things can be true at once. I can mourn for what might have been, and also be thankful for what is.”

cystinosis has made me more health conscious. I try to control the things I can. I think I eat healthier and get more activity than if I didn't have it. The discipline required to manage

cystinosis has benefited other areas of my life. I believe in the paradox that two things can be true at once. I can mourn for what might have been and also be thankful for what is.

Global Genes RARE Concierge Patient Services

What is RARE Concierge Patient Services?

Global Genes' RARE Concierge patient service guides are here to help you navigate the complicated world of rare and undiagnosed diseases. No matter where you are in the world, your journey, or what your unique need is, RARE Concierge can help you.

The team has decades of personal and professional experience in rare disease advocacy and support. As patients, caregivers, and advocates, we understand the challenges you and your family face.

RARE Concierge can provide resources, education, and important connection free of charge, including:

- * Finding a specialist or care resources
- * Understanding research or clinical trials in your disease
- * Locating social, emotional, or mental support.

Contact a patient services guide at <https://globalgenes.tfaforms.net/5029224>.



Now Recruiting - Novartis CYStem Phase I/II Clinical Trial in Cystinosis

Novartis is running an open-label, multi-center, CYStem phase I/II study to assess the safety, tolerability and efficacy of DFT383 in pediatric participants with nephropathic cystinosis, followed by a long-term extension phase. DFT383 is an autologous hematopoietic stem cell (HSC) gene therapy.

The purpose of this clinical study is to assess safety, tolerability, and efficacy of DFT383 in nephropathic cystinosis participants 2 to 5 years old. The study includes two treatment groups (Cohort 1 and Cohort 0) and consists of a Core Phase and a long-term Extension Phase. Participants in Cohort 1 will receive DFT383 and participate in both the Core and Extension Phase. Participants in Cohort 0 will not receive study treatment and will participate in the Core Phase only. The two cohorts will be run in parallel. Investigational sites may participate in one or both cohorts (see below).

Cohort 1: Approximately 15 participants will receive treatment with DFT383 in 3 (sub) cohorts (1A, 1B and 1C) dosed in a staggered approach. The total study duration for a participant in Cohort 1 will be up to 32 months in the core phase and up to 13 years for the long-term extension phase.

Cohort 0: Approximately 15 participants meeting similar inclusion/exclusion criteria and receiving Standard of Care (SoC) will be enrolled. The Schedule of Activities will be reduced for this Cohort. Cohort 0 is not a direct control but will provide essential context for interpreting the results observed in the participants receiving DFT383. The total study duration for a participant in Cohort 0 will be up to 24 months.

Further information such as inclusion/exclusion criteria is available at clinicaltrials.gov (scan the QR code) and through Novartis Pharmaceuticals at **1-888-669-6682** or novartis.email@novartis.com.



CYStem Study Sites

Georgia

Children's Healthcare of Atlanta - Emory (Cohort 0 only)
Contact & Principal Investigator: Larry Greenbaum
404-712-6374 lgreen6@emory.edu

California

UCSD - Rady Children's Hospital
Contact & Principal Investigator: Caitlin Carter
858-966-4953 cecarter@health.ucsd.edu

Texas

Baylor Texas Children's Hospital, Houston (Cohort 0 only)
Contact & Principal Investigator: Ewa Elenberg
832-824-3800 elenberg@bcm.edu

Stanford University - Children's Health

Contact: Research Nurse
650-725-9032
scgt_clinical_trials_office@lists.stanford.edu
Principal Investigator : Alice Bertaina

Education & Awareness Update

By Marybeth Kruppenacker, Vice President of Education & Awareness



Laura just turned 40 years old, and these are some of the important people in her life, Dr. William Gahl, Dr. Rick Kaskel, Dr. James Drukakis, and Maya Doyle. A rare disease takes a team. Laura has been blessed in part because of this amazing team.

Wow, Laura's 40th birthday just passed. As I re-read my Education and Awareness article from last spring, another year has come and gone and we are truly amazed and blessed!

I had posted a photo on Laura's 40th birthday with some of her "team" members who have, and continue to have, a profound effect on her life these last 37 years. There have been many more doctors and various professionals who made Laura's 40th birthday special, it truly does take a village! I am so very grateful to all of those who called, sent a card or a note wishing her well. She had a wonderful weekend with friends and family and thoroughly enjoyed each special moment. We are both blessed.

The Cystinosis Research Network is certainly one of our biggest blessings. We continually explore different ways to be part of the rare disease community as a whole and be a

voice. We have spent the last several years attending and exhibiting at professional meetings throughout the country. CRN's representatives have become the face of cystinosis through education and raising awareness at these events. While these meetings have been wonderful, we've found the exposure and exhibit times have been cut dramatically. We have decided to shift our approach by increasing virtual engagement, exploring the expansion of regional gatherings, and investigating additional professional forums to collaborate with. What I have learned over the last 37 years is to ask questions. The world of kidney transplant is rapidly changing, as is rare disease research. The more you can arm yourself with information and ask questions, the more you understand.

Life is moving fast, and I believe in the world of medical research it is moving faster than ever. 37 years ago, I had

no idea or understood what a rare disease was, and being handed that diagnosis, all I could do was to focus and pray for Laura to turn four years old. Never did I think we would get to 40! But she has, and continues to live life to the fullest, surrounded by love and a positive joyful outlook that has carried both of us forward. I recently read something I will share. "It is not joyful people who are grateful, it is grateful people who are joyful". We are grateful for all that the world of cystinosis has brought into our lives.

One powerful way our community comes together each year is through Cystinosis Awareness Day. Learn how you can be a part of this year's campaign on [Page 24](#).



The Live Like Laura Fun Fund was established in memory of Laura McGinnis, to allow those who live with cystinosis to experience life's adventures and fun as Laura loved to do. Learn more about the fund and how to apply at cystinosis.org/llff.

My Chicago Dream and the LLLFF

By JB Shepperd

Despite only interacting with Laura a couple of times during my childhood, I have always considered her to be an important person in my life. My family never had much interaction with other cystinosis families, outside of a conference we attended once, and the few times that I got to see Laura. We were both very young at the time, probably around 10 years old, but I remember connecting with her almost immediately and having a great time just being kids together. As I got older and my understanding of cystinosis grew, it was very easy to feel isolated. Nobody around me really knew what my sister and I (who is also a cystinosis patient) were going through. In these moments, I would often think of my time with Laura. Knowing that there is someone else out there going through the same things, taking the same meds, and dealing with the same social difficulties (namely the smell associated with the medication) was comforting. Even if she was living across the country and we weren't directly in touch, I wasn't alone.

I believe a similar feeling of isolation is what drove me to attend the CRN conference at Grand Rapids in 2025. In my adulthood, I felt disconnected from the cystinosis community. I was unhappy with my living situation, worried for my future and felt just generally lost. It was there that I reconnected with Laura's mom, Frankie. She recognized me



Childhood memories, JB in the Cowboy hat and family with Laura McGinnis far right.

immediately after I walked in on the first day of the conference. It was through Frankie that I learned about the Live Like Laura Fun Fund. I had plans to visit Chicago immediately after the conference, mostly to see if I liked the city enough to move there. Cystinosis has developed in my eyes to the point where I don't entirely feel comfortable driving, so I knew that if I wanted to pursue real independence, I would need to be somewhere with public transit. After learning of the LLLFF, my chances of being able to move to somewhere like Chicago were suddenly much higher than before. So, I went to Chicago, thinking of Laura and how her legacy could help to bring me a level of freedom I had never experienced before.

And I absolutely loved it! The city was beautiful, the food was amazing and the people were so incredibly social. I felt like I had finally found somewhere I could call home for the rest of my life.

I skipped all the 'tourist-y' attractions because I knew that with the help of the LLLFF and some hard work, this could be my home forever. I walked through various neighborhoods, with an almost perpetual smile plastered on my face, as I considered which of these areas I would move into. I'll never forget how that felt. I knew this city was where I needed to be, where I could build a life for myself that I would be truly happy and free. No more feeling locked inside for days at a time because I couldn't drive. No more paying ridiculous amounts of money for rideshares just so I could spend time with friends or make new ones. Now, I can just hop on a train/bus and be virtually anywhere!

When I returned home, I started the moving process immediately. My family was a bit jarred, but I knew I wanted to move as soon as possible. I applied for the LLLFF, and once I was accepted, I used that money to make

My Chicago Dream and the LLLFF, *continued*

the move possible. Within as little as a month, I was on a plane with my (very anxious) cat in one arm, and all of my essential belongings stuffed in a suitcase that barely closed, ready for my new life. Now, here I am in Chicago six months later, loving life more than ever. I'm fully settled in with an apartment I love in a neighborhood that feels like home, and even a few new friends to show me around the city. I still have moments where I walk down the street, look around, and

have to remind myself that I really do live here. It is truly a dream come true. Without Laura's wonderful legacy in the form of the LLLFF, I would still be stuck in Texas at this moment, alone and trapped in my own home.







So please, if you are a patient with cystinosis or sibling to someone with cystinosis, take the leap and apply for the Live Like Laura Fun Fund. Go on that trip, buy those concert tickets, do something that makes you or your

child/sibling happy. Cystinosis did not rule Laura's life. For a long time, it felt like cystinosis was ruling mine. Don't let it rule yours or your child's. From the bottom of my heart, thank you to Frankie for creating this fund. Thank you to CRN for helping maintain it, and thank you to Laura for being you. I hope to continue to live like, Laura, and break down any and all roadblocks to be the happiest I can possibly be.

What's Your 57?

In celebration of the 9th Annual Cystinosis Awareness Day on May 7, we are asking the community, "What's your 57?" It is a call to action that can be big or small. We encourage everyone to choose something meaningful to them. If comfortable, share with neighbors, friends, loved ones, healthcare professionals, anyone who will listen. These simple actions can help others better understand cystinosis, drive earlier diagnosis and better care, generate stronger support, all in hopes of improving long-term outcomes.

Here are a few ideas on how to take part:

-  Reflect on your cystinosis journey and write 57 words on how you are feeling today (share or keep it private)
-  Create a 57 second explanation about cystinosis to bring up in conversation
-  Spend 57 minutes reading the latest on cystinosis and share one takeaway
-  Ask 57 friends/family to donate to the CRN
-  Consciously perform 5-7 acts of kindness
-  Text, email or call 5-7 people to talk about awareness day; tell them why it matters to you



9th Annual
**Cystinosis
Awareness
Day**
MAY 7, 2026

How will you choose to honor this momentous occasion? Share with us at info@cystinosis.org or tag us on social media.

It's not too late to donate!

Use our secure, tax-deductible form at <https://tinyurl.com/donate4CAD>.

Development Update

By Jonathan Dicks, President / Vice President Development



Jon, Jeff, and Ella taking a break from their advocacy work on The Hill.

As we move through another year together, I find myself reflecting on the incredible strength of this community. Time and again, we see what is possible when families, patients, clinicians, researchers, and advocates unite around a shared mission. The progress we've made—whether through advancing research, expanding support programs, or strengthening advocacy—has always been fueled by one powerful force: each other.

That spirit of connection has always been the heartbeat of the Cystinosis Research Network. From the earliest days of this organization, families and supporters stepped forward not only to seek answers, but to help create them. They raised awareness in their hometowns, organized gatherings with friends and neighbors, and shared their stories in ways that opened doors for research and hope.

Today, that same spirit continues to drive meaningful progress. We are encouraged by the continued strength of our partnerships across

the pharma and biotechnology community. Because CRN has long worked alongside scientists, clinicians, and industry leaders to help shape priorities in cystinosis research and care, we are pleased to share that we remain on schedule with strong industry support for the work ahead. These collaborations reinforce CRN's long-standing commitment to accelerating the discovery of a cure, improving treatments, and enhancing quality of life for those living with cystinosis—ensuring that important scientific progress continues while our community remains at the center of that effort.

As we look ahead, one of our greatest opportunities lies in continuing to grow the grassroots energy that has always defined this community. CRN is here to support those efforts. Whether someone is planning a small local gathering, a school fundraiser, a community walk, or an online campaign, we want to make it easier than ever for members of our community to take action. Even the simplest idea—bringing people together around a shared cause—can grow into something meaningful. And for those who want to support the mission in a direct way, donations can always be made through the CRN website at www.cystinosis.org, where every contribution helps sustain the programs and research that serve our families.

This year we are also launching a new initiative designed to inspire participation across our entire community: #WhatsYour57?

For many in our community, the number 57 carries deep scientific

significance. The 57 kilobase deletion is the most common genetic mutation associated with cystinosis. While it represents a critical part of the biology behind the disease, we also see it as an opportunity to transform that number into a symbol of action and resilience.

The idea behind #WhatsYour57? is simple. "Your moment. Your pace. However you show up, find your 57."

Maybe it's 57 minutes spent advocating or raising awareness. Maybe it's 57 seconds to share your story online. Maybe it's 57 daily deep breaths as a reminder to care for yourself while caring for others. For some it may mean a physical challenge, like 57 push-ups or a 57-minute walk with friends. For others it might be making a \$57 donation, or encouraging 57 people to learn about rare disease.

The goal is not perfection or scale. The goal is participation. Small moments of action, multiplied across an entire community, become something powerful.

Family conferences will always be a cornerstone of the cystinosis community. They provide moments of learning, connection, and renewal that remind us why this work matters so deeply. But the years between conferences are just as important. Those are the years when our mission spreads outward—into schools, neighborhoods, workplaces, and communities around the world.

That's where grassroots energy becomes transformative.

Imagine dozens of families hosting events in their own communities

Development Update, *continued*

during an off-conference year. Imagine classrooms learning about rare disease for the first time, neighbors rallying around a local fundraiser, and new supporters discovering the mission that has united us for decades. Each conversation builds awareness. Each event builds momentum. Each act of generosity brings us closer to better treatments and, ultimately, a cure.

Cystinosis may be rare, but the strength of this community is extraordinary. Every story shared, every fundraiser organized, every act

of support helps carry the mission forward.

If CRN has made a difference in your life—or in the life of someone you love—I invite you to be part of this next chapter. Share your story. Join the #WhatsYour57 movement. Organize a community fundraiser. Or support the mission with a donation through www.cystinosis.org.

Together, we continue to prove what has always been true: when this community moves forward together, progress follows.



Jonathan Dicks with Ron Bartek, Co-founder & President of Friedreich's Ataxia Research Alliance.

CRN Offers Memorial Fund

By Karen Gledhill, Secretary

The Cystinosis Memorial Fund was created to give individuals with cystinosis the opportunity to receive a grant of up to \$1,000 USD. The grant could be used to pay for a range of expenses from a college class to a new software program or a meal plan at school. We've expanded to include muscle wasting prevention programs, exercise programs, nutrition counseling, yoga, even home exercise equipment.

We have been pleased to award various grants in past years and hope you will consider applying. If you are interested, go to <https://cystinosis.org/cmf>.

Cystinosis MEMORIAL FUND

Learn more at
cystinosis.org/cmf

Family Support Update

By Chelsea Meschke, Vice President of Family Support

We're so excited to continue our journey with this amazing community.

I truly love being part of the Cystinosis Research Network and witnessing all the incredible work being done to support and uplift families like ours. From participating in conferences across the globe to learn about new resources and ongoing research, to being an essential and integrated part of that research itself, this community continually shows up to support the doctors, researchers, and families who make progress possible.

Living in the rare disease world introduces us to a lot of terminology that helps describe different aspects of our journey. These concepts are important not only to recognize, but also to use as tools to better understand our experiences.

One of these concepts is peer support networks. These include our online forums and social media groups, patient-led nonprofits, and the direct support from families who have come before us and now serve as mentors in many ways. These



Cystinosis warriors, Jaxon and Myles Meschke.



The Meschke family, Chelsea, Myles, Brian and Jaxon.

networks provide deeply meaningful emotional support during some of the most pivotal moments in our journeys. These peers are the ones who help guide us through the dark moments, celebrate our victories, and carry us through the lows. It's incredible how the kindness and love of what once felt like "strangers" can grow into the love and embrace of family.

Another major concept in the rare disease world is called hope cycles. This idea describes the emotional ups and downs that rare disease communities often experience. Optimism and disappointment can revolve around many aspects of living with a rare illness. Clinical trials, new research breakthroughs, and experimental treatments can spark tremendous hope and excitement about the possibility of a brighter future. At the same time, delays in timelines, limited funding, or the challenges of navigating regulatory processes can bring disappointment and frustration. These cycles of hope can sometimes be just as emotionally draining—if not more so—than the day-to-day realities of living with a rare disease.

My purpose in sharing these concepts is simple: to help name some of the feelings many of us experience, recognize that they are real, and remind us that we can take back the power that sometimes feels taken from us. We are not alone in this rare world. While no two cystinosis patients are exactly the same, we are walking this journey together, and I hope that knowledge brings comfort.

I am truly honored to continue serving as your Vice President of Family Support, and I look forward to everything ahead for our community.

In May, we will be gathering in Jacksonville, Florida for a regional family meet-up. It will be a one-day event, but it will be filled with connection, learning, and support.

Please remember that if you ever have questions, need support, or simply need a shoulder to lean on, we at CRN are always here for you.

Be sure to watch our social media pages for upcoming Zoom support meetings!

For any needs or inquiries, please feel free to contact Chelsea Meschke at chelseam@cystinosis.org.



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Carson's Story

By Emily Hughes

Our family lives near Jacksonville, Florida. We have a 4-year-old daughter, Adalyn, and Carson, who will be two in May. Carson was born weighing 7 lbs 15 oz, and we had no concerns leaving the hospital with him. He had a "normal" first year of life, and we had very minor concerns. Around 11 months of age, he started to refuse to eat foods. We thought he was just a distracted eater, as our house is always busy, and he would rather play than sit in a high chair and eat. He dropped from the 50th percentile to 37th in just a couple weeks. This didn't alarm his pediatrician yet, as this was the same time he started walking and he was extremely active.

Around 12 months of age he started wanting to drink an excessive amount of water. We were in and out of the pediatrician's office checking his weight and asking for blood sugar checks, because we thought he could have type 1 diabetes. His blood sugar continued to come back normal and his weight continued to drop.



Carson and his Adalyn.



The Hughes family having fun with Mickey Mouse.

We finally took him to the emergency room to get labs, and to get him checked out overall. They admitted us to the local children's hospital for 15 days for failure to thrive. There, we found out he had Fanconi syndrome. They did three procedures under anesthesia: a GI scope, a kidney biopsy, and a g-tube placement. His kidney biopsy came back normal, throwing our local nephrologist for a loop. We were told to be seen as an outpatient, despite desperate requests for answers. Our local nephrologist sent us to multiple specialists, and told us we were likely "one in a million" with a case of Monoclonal Gammopathy of Renal Significance (MGRS). This is typically only seen in older adults. In his 55+ years of practice the doctor had seen a transient case Fanconi syndrome one time.

We left upset and wanted a second opinion. We reached out to Dr. Shoemaker in Gainesville via email in a desperate attempt to find answers for Carson. He and his team worked magic and got us in within two days. He suggested possible Dent Disease based on his growth and appearance, but performed the Natera test to see

for sure in the office office. We got our results back exactly three weeks later with the diagnosis of cystinosis.

It's been a long and difficult year, as we have had to navigate and overcome the challenges cystinosis brings. He is on around-the clock medications and supplements that are becoming our new routine. While this is not easy or fun, we try to make it a little lighter by singing a song, or having him help push his medication through the g-tube. He has an amazing big sister who loves to help with this too.

In January, we had the privilege of meeting Dr. Gahl and his team at the NIH in DC. Carson spent two days getting tests done, seeing specialists, etc. before we sat down with Dr. Gahl to review on the third day. We are grateful for our experience, and hopeful others can experience the same. We are also thankful for Cystinosis Research Network and their support throughout our journey. As a family, we look forward to continuing to support the CRN and raise awareness of cystinosis in our community.

- Daniel, Emily, Adalyn, and Carson Hughes

The Moms' Trip

By Frankie McGinnis, Director

Over the last 25-plus years that I have been attending Cystinosis Conferences, the talk among the moms always leads to the idea of taking a trip. The idea of a carefree weekend, no one to think about other than yourself, no schedules, no timers was always the dream. Carefree...not a word that is related to motherhood in general, much less motherhood that involves people with high medical needs. As mothers, we naturally put everyone else's needs ahead of our own; it simply becomes part of the role. As the years went on, so too did the talks of the mythical getaway.

I have traveled extensively with some of the other cystinosis moms, but it has been wrapped in other obligations, conferences, fundraising, advocating. Never just to dwell in the understanding of those that "get" living the life of a caregiver/mother. Don't get me wrong, I would not trade the 23 years that I had with Laura for anything. Being her mom was a privilege. What I will say is that worry is at another level when you wonder, will your person need a major operation? Am I managing things so well that they stay as healthy as they can be? When I talk with the mom in the neighborhood who is worried if Billy will get a cold; well it is just not the same. This year at the conference in Grand Rapids, the talk of the mythical trip ensued again. A few of us were comparing notes, their people were doing well, no looming 'monster' on the horizon. Could this trip be a possibility? We all went home with dreams of umbrella drinks in our minds.



Frankie, Julie and Stephanie enjoying the sites in Costa Rica on their recent 'mom' getaway.



Stephanie Hassan, Frankie McGinnis, and Julie Ayers.

The Moms' Trip, *continued*

A few weeks after getting home as I sat at work, I sent a text message. Could the trip be a reality? The excitement that I was met with was palatable, and by the end of a few hours the trip was completely booked. Fast forward five months and we were heading to Costa Rica! Sunshine in the cold months was just what these women needed, add in the talks, the laughs, the tears, the love- well, priceless is an understatement. We ate, drank and were merry and the bonds formed over all those years of life together were further cemented. When Laura died, I was not sure

where I would fit in this community. What I realized with the last two conferences and this trip was if you choose, regardless of what happens in life, this community is here.

I would love to plan a trip for moms every year or at least in non-conference years. A chance to just be. I hope more people will join in the future, and this can be part of the self-care everyone in a caregiving role needs to do. If this sounds like what you would like, reach out- maybe we can be the catalyst that fuels this idea. Moms taking care of Moms.



Julie, Stephanie, and Frankie taking in the nightlife in Costa Rica.

CFUK Celebrates £3.9 Million Award to Advance CF10 Toward Clinical Trials

By Alex Hutchison

The Cystinosis Foundation UK (CFUK) announced a major milestone: researchers at the University of Sunderland received £3.9 million from the UK Medical Research Council (MRC) to advance a promising new treatment, CF10, into clinical trials.

CF10, a novel prodrug, was discovered by the late Professor Roz Anderson, whose research aimed to improve therapies for cystinosis. Current treatment, cysteamine,

requires frequent dosing and often causes severe side effects such as nausea, ulcers, and body odor. CF10 is designed to deliver therapy more directly to affected cells, potentially reducing doses and side effects, with the goal of improving quality of life for patients and families.

This clinical trial builds on earlier support. In 2018, CFUK's efforts helped secure £1.6 million in MRC funding for preclinical development.

Now, the trial will be led by Professor Herbie Newell at Sunderland, with partnerships including High Force Research (Durham) for drug manufacturing, Newcastle Specials for capsule production, University Hospitals Birmingham NHS Foundation Trust for trial management, and Birmingham Women's and Children's NHS Trust and CEMAS (Wokingham) for sample analysis. Dr. Graham Lipkin, kidney specialist and cystinosis expert, will oversee dosing studies.

This funding marks a critical step toward evaluating CF10 in patients and highlights the combined efforts of scientists, clinicians, CFUK, and families. If successful, CF10 could transform treatment for cystinosis worldwide.

Adapted with permission from CFUK.

“*CF10 could change everything. This isn't just a scientific milestone— it's hope for a brighter future.*”

- Will Newman, CFUK Chair

Good News

Editor's note: In this issue of our Good News section we are featuring Laura Kruppenacker, honoring her cystinosis journey as she recently celebrated her milestone 40th birthday. We are also featuring our newest CRN Board member, Hannah Creel, celebrating her recent engagement to Jon Logan. We send our congratulations to both of these wonderful members of our cystinosis community.

Forty and Fearless: Laura Kruppenacker



Laura Kruppenacker celebrating her milestone 40th birthday.



Laura celebrated her 40th birthday with her friends, and family, enjoying a spa weekend.



Aunt Laura with her nephew Finley and niece Parker.



Aunt Laura with Finley and Parker and their friend Coach.



Laura with her godson, Tyler.

Good News

Hannah Creel & Jon Logan Beadlescomb Engagement

By Hannah Creel, Director

Hannah Creel and Jon Logan are delighted to announce their engagement after a love story that began with a simple coffee date in Birmingham, Alabama. For their first date, Jon drove nearly an hour to meet Hannah at a local coffee shop, and what began as a quick cup of coffee soon turned into lunch, thrift shopping and a walk through a nearby park. By the end of the day, their first date had lasted nearly eight hours—and they have been together ever since. This spring, the couple will celebrate two years together, a relationship Hannah says was defined from the beginning by Jon Logan’s warmth, compassion, and genuine care for others.

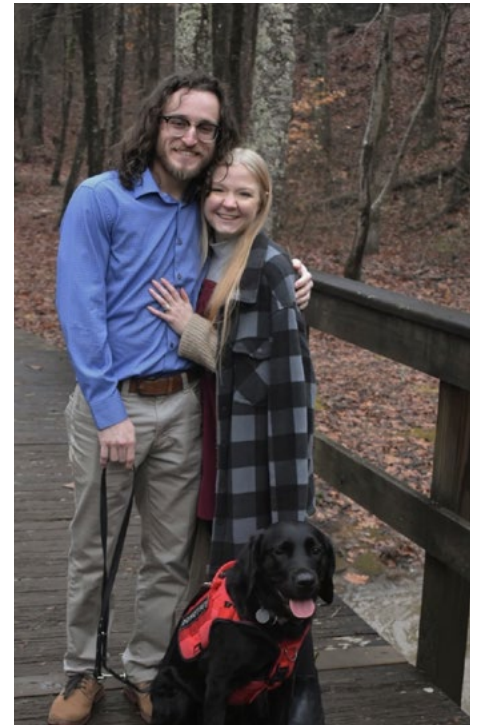
On Sunday, January 25, 2026, Jon proposed during a walk in the very same park where their first date ended. Earlier, when he asked Hannah’s parents for their blessing,



Jon Logan and Hannah.

he unknowingly shared a date with deep meaning: January 25th was the day Hannah’s parents had learned their IVF treatment was successful, and that they were expecting her. Overwhelmed by the significance of the moment, the couple returned to the park after church as they often do. With Hannah’s service dog, Hope, happily distracting her, Jon Logan called her name—when she turned around, he was down on one knee. True to her easily spunky personality, Hannah responded with an excited scream of joy before saying yes.

The couple is thrilled to be planning their wedding for Saturday, July 18, where they will celebrate the beginning of their life together—with Hope faithfully by their side. From the very beginning of their relationship, Jon has been a steadfast source of love and support in Hannah’s journey with cystinosis. His patience, compassion, and willingness to walk alongside her through the challenges the disease can bring have been a constant encouragement. Together, they share a passion for advocacy and for empowering others within the cystinosis community, including their involvement with the Cystinosis Research Network. Hannah is grateful



Jon Logan, Hannah Creel and their companion, service dog Hope.

to love and be loved by someone who is not frightened by the difficulties of cystinosis, but rather someone who sees her value and worth. Someone who sees the beauty in this cystinosis journey and community. Everyone deserves to be loved in this way, and cystinosis should not rob anyone of that opportunity should they choose to have it.



CYSTINOSIS SOLIDARITY
CNE INTERNATIONAL CONFERENCE 2026
CYSTINOSIS IRELAND AND CYSTINOSIS NETWORK EUROPE

HAVE YOU REGISTERED?

The CNE International Conference will be hosted by Cystinosis Ireland in Dublin in July. What can you look forward to?

We are offering

- Supervised childcare and activities for children up to 12 years old
- Translation from English to Dutch, French, German, Italian, Spanish, Turkish, Russian
- Specific activities for teenagers and adults with cystinosis
- A chance to meet and connect with peers, healthcare professionals and researchers, all experts in cystinosis

Agenda highlights include:

Thursday 2nd July

- Family welcome evening and dinner
- Closed meet and mingle cocktail hour for adults with cystinosis and their partners

Friday 3rd July

- Introduction to cystinosis
- Breakout discussion groups by age and experience of cystinosis
- Research updates on key projects of interest to the community
- Annual research prize award for early stage career researchers

Saturday 4th July

- Artificial Intelligence in research and what that means for cystinosis
- Themed breakout sessions
- The importance of clinical guidelines and multidisciplinary clinics
- Updates from international cystinosis organisations
- A chance to give your input to future research priorities
- Closing dinner and celebration

www.cystinosis-europe.eu/our-conference

“I tell my sons cystinosis is something you have, it’s not who you are.”



I choose Cystadrops because...

- It dissolves crystals across all layers of the cornea
- Provides flexibility of 4x a day dosing
- The CystaTouch dropper is easy to use

– Nicole, mother to three teenagers living with cystinosis

Indications and Usage

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

Important Safety Information

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

Visit www.cystadrops.com for full prescribing information



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www.recordatirarediseases.com/us PP-CDPS-US-0245



PATIENT BRIEF SUMMARY

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

Summary:

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

What is Cystadrops?

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

What should I know about using Cystadrops?

- To minimize the risk of contamination, do not touch the dropper tip to any surface. Keep bottle tightly closed when not in use.
- Cystadrops contains the preservative benzalkonium chloride. Direct contact with soft contact lenses should be avoided. Remove contact lenses prior to application. Lenses may be reinserted 15 minutes following administration.
- A condition where the pressure inside the skull increases for unknown reasons has been reported with cysteamine taken by mouth or cysteamine eye drops (used at the same time as cysteamine taken by mouth). This condition went away with the addition of medicine that increases the production of urine.

Before you use Cystadrops, tell your doctor if you:

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

How should I use Cystadrops?

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

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

What are the possible side effects of Cystadrops?

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

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

How should I store Cystadrops?

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

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

General information about the safe and effective use of Cystadrops.

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

What are the ingredients in Cystadrops?

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

Preservative: benzalkonium chloride 0.1 mg/mL;

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

Manufactured by: Baccinex SA, 2822 Courroux, Switzerland

Manufactured for: Recordati Rare Diseases Inc., Bridgewater, NJ 08807, U.S.A

**For current prescribing information, please visit
www.CYSTADROPS.com/PI.**

Revised: July 2024

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Personalized one-on-one support is available for patients receiving treatment with CYSTADROPS and their caregivers. Dosing and administration information along with additional resources are within easy reach. The PL program is also available in Spanish. Enrollment is voluntary.

*Information provided by the PL program is for educational purposes only and is not intended to replace the advice of patients' healthcare providers.

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



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Research Update

By Kristina Sevel, Vice President of Research

As a parent advocate and Vice President of Research at the Cystinosis Research Network, this work is deeply personal to me. We live every day with both the challenges of cystinosis and the hope that research brings, and I am so inspired by the dedication of our research community and the strength of the families who drive this work forward.

Because of your support, researchers are pushing forward—learning more, asking smarter questions, and building on progress that truly matters to our community. In the updates that follow, you'll hear directly from these investigators in clear, plain language about what their studies are, and why it's important.

Every study represents hope: hope for better treatments, fewer burdens, and brighter futures for our children and loved ones. Continued support from our community makes this progress possible. If you are able,

please consider supporting cystinosis research so that together we can keep this momentum moving forward and

bring new possibilities to families living with cystinosis at tinyurl.com/CystDonate.



Mike, Grace and Kristina visit Santa.



Kristina and Grace decorating a gingerbread house.



The Sevel family, Mike, Grace and Kristina.

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

By Stefanie Witt, Katharina Hohenfellner, and Julia Quitmann

The aim of the QUALIFY study was to develop and validate a questionnaire to measure health-related quality of life among children, adolescents, and young adults living with cystinosis. Having a validated, disease-specific questionnaire is an important step toward improving patient-centered care and evaluating new treatments or therapies.

Over the past years, we have worked closely with many families to better understand what matters most in everyday life with cystinosis. Their experiences and perspectives helped us identify the key areas that influence quality of life. Based on this valuable input, we created initial questionnaire statements and carefully refined them after a pilot test. The revised version was then tested in a larger field study and a retest phase to ensure the questionnaire is reliable and scientifically sound. As a result, the final version of the QUALIFY questionnaire is now available.

QUALIFY measures health-related quality of life in children, adolescents, and young adults with cystinosis using age-appropriate versions. There are two caregiver (proxy) versions for families of children aged 0–7 years and 8–26 years, as well as a self-report version for young people aged 8–26 years. The questionnaire covers key areas of daily life, including physical wellbeing, emotional wellbeing, social wellbeing, and experiences with medical care. For older age groups, it also includes questions

about future perspectives.

All versions of QUALIFY are based on the same overall concept but are adapted in length and detail to match the child's age and developmental stage. The questionnaire is currently available in German, English, Spanish, and French. While the German and English versions have been thoroughly tested, more families are still needed to further validate the French and Spanish versions.

We are deeply grateful to all families who have already contributed to this important work. Their participation helps us better understand life with cystinosis and supports the development of more patient-centered care in the future.

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

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

Male infertility is a significant but poorly understood complication of cystinosis. To better understand

why this happens, Dr. James Smith, Director of Male Reproductive Health at UCSF, Dr. Minnie Sarwal, transplant nephrologist and cystinosis expert, and Dr. Sur, cystinosis researcher at UCSF, are leading a study examining the causes of infertility from adult men with nephropathic cystinosis, comparing also for overlapping and unique causes of infertility in men without cystinosis and healthy men with normal fertility.

These studies have helped to identify the biological changes that may be affecting overall reproductive health and specifically in the case of cystinosis. The team have also discovered that a natural compound may help protect affected gonadal cells in cystinosis and the team is now working on further developments for as a potential treatment for male infertility. This work has been funded by the HRB and the Cystinosis Foundation of Ireland

Cognitive Control Systems in Cystinosis

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

Research has shown that individuals living with cystinosis may experience challenges with cognitive function, including how they process and remember information. Early studies have indicated some differences in behavior and brain function in people with cystinosis, particularly in areas like sensory memory. However, we still don't fully understand how cystinosis impacts brain development

Development of a patient-reported outcome, *continued*

and cognitive abilities over time, and there's much more to learn about the specific areas where individuals with cystinosis may face difficulties. Why is this important? By identifying the areas of cognitive vulnerability in cystinosis, researchers can work toward developing therapies and strategies to support individuals in overcoming these challenges and improving their quality of life. To help achieve this, our team is focusing on understanding key aspects of executive functioning, which include

skills like memory, managing tasks, adapting to new situations, and handling conflicting information. To dive deeper into this, we're conducting a study using advanced non-invasive technology called high-density EEG, which measures brain activity in real time, along with standardized cognitive assessments. We'll be testing children, teens, and adults with cystinosis, as well as individuals without cystinosis for comparison. Additionally, we'll examine the role of genetic mutations versus the disease





itself by studying individuals who carry the gene for cystinosis but don't have the disease. This research is inspired by previous findings, existing knowledge about cognitive challenges in cystinosis, and personal insights shared by patients and families. By better understanding how cystinosis affects the brain and cognition, we hope to pave the way for new solutions that can make a meaningful difference in the lives of those impacted by this rare condition.

CYSTINOSIS WARRIOR IMPACT PROGRAM



The Cystinosis Warrior Impact Program (CWIP) was designed with an ambitious goal: to positively impact each person living with cystinosis through mental health support, financial assistance (scholarships), medical resources (support programs), education, or connection to critical networks. We encourage you to reach out today to learn how CWIP can support you or someone you know.

How to get help or get involved:

-  **Contact us.** Email info@cystinosis.org and we can confidentially discuss how to support your needs.
-  **Referrals.** Do you know a cystinosis warrior in need? Provide them with our email to get started.
-  **Resources.** Know of a great program or resource the cystinosis community qualifies for? Send it our way and we can expand the cystinosis resource library.
-  **Share.** Help us spread the word to reach even more cystinosis warriors.

“ Thank you for being a source of comfort. ”

“ It literally doesn't matter what I need, I know I can call on you at any time. I still remember our first conference spent together in Philadelphia. The CRN has been with us every step of the way. ”

“ THANK YOU for introducing us to this doctor, he has been such a dream to our family and [our daughter's] health. ”

“ Thank you to the Cystinosis Research Network for making the connection. These moments matter. And to anyone in the community who has a story to tell - share it, however you can. Your voice could change someone's path - or even save a life. ”

Financial Update

By Frank Re, Treasurer

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 2025 for the Family Conference held in July, in beautiful

Grand Rapids Michigan, which brought together hundreds in the cystinosis community along with both medical and industry partners. CRN's net income for calendar year 2025 was \$32,449. 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, research grants and scholarships. That being said, positive net income for 2025 exceeded our budget, thanks to the tireless efforts

of your Board members, especially President and VP of Development, Jonathan Dicks.

Thanks to grants and fundraising, CRN's current equity (assets minus liability as of December 31, 2025) stood at roughly \$622,000, which is critical in funding additional research as well as our next Family Conference in 2027. 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.



Care Package Program

"Our son, Carson, was diagnosed with Cystinosis in August. This was a devastating and difficult time for our entire family. We received a care package soon after from the Cystinosis Research Network that had lots of things to help with the new challenges we were facing. It included a water bottle, sensory toys, and other goodies to make our adjustment a little easier. We were so appreciative to have something positive sent to us during a challenging time for our family."

- Emily

[CYSTINOSIS.ORG/CARE-PACKAGE](https://cystinosis.org/care-package)

When life with cystinosis feels overwhelming, even the smallest gestures can make a difference. The Care Package Program was created to remind families and individuals that they are not alone on this journey. Each package is thoughtfully assembled with supportive materials. On the next page, you'll find details on how to receive a package for yourself or nominate someone you care about.



Carson Hughes.

2025 Donor Honor Roll

\$100,000+

Amgen, Inc.
Leadiant Biosciences, Inc.
Recordati Rare Diseases Inc.

\$50,000-\$20,000

Novartis Pharma AG
Magistro Family Foundation

\$7,500-\$2,000

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Manning, Lawrence
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\$1,999-\$1,000

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Wood, Kathy
Thompson, Baylie
Palmer, Tanya
Parker-Thomas, Alichia
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Visakan, Sasivathany
Kumar, Pavan
Prasadh, Anitha
Thangarajan, Sumathi
Mitchell, Sabra
Meta
Morris, Jan
Langley, Edward & Joyce
Ryczek, Rick & Kristi

Help Support the CRN's Mission – Donate Today!

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

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

We take great pride in carrying out our motto: "Dedicated to a Cure. Committed to our Community"

We are here to for you, whether you are:

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

The Cystinosis Research Network is proud to provide valuable resources to the community, free of charge. Many can be found on our website: cystinosis.org.

Resources include but are not limited to:

- The latest cystinosis information through our biannual CRN Newsletter; The Cystinosis Advocate, our website (cystinosis.org), the popular online cystinosis Facebook support groups, regular email updates and other social media channels.
- CRN Family Conferences and Regional Meet Ups. Exchange knowledge and create friendships with other families and individuals living with cystinosis. Learn first-hand the latest discoveries about cystinosis from medical professionals.
- Rare Disease Week Scholarships. Participate in a week-long event in Washington, D.C. Let your voice be heard by legislators and policymakers who need to know why cystinosis (and other rare diseases) is important to you.
- Access to Cystinosis Research Network individuals and families near you.

Please consider donating to the CRN today to help us continue offering robust support, education and research to the global community.

Ways to donate:

- Scan the QR code using your mobile camera
- Use the envelope provided to contribute via check
- Visit our website donation page at <https://tinyurl.com/CystDonate>



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

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CYSTINOSIS
RESEARCH NETWORK

Volume 18, Issue 1
Spring 2026

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Editor Terri Schleuder

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

CRN VISION

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

CRN MISSION

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