



THE CYSTINOSIS Advocate

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2023 CRN Family Conference *Nashville, TN*

The 11th biennial conference is a chance for our worldwide cystinosis community to come together. After years apart, we hope you join us to rekindle old friendships, make new connections, and learn from and speak with clinicians experienced in treating and researching cystinosis.

Save the Date
July 13 - July 15

Nashville Marriott
at Vanderbilt University

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The President's Letter

Since our last newsletter, the Cystinosis Research Network has been humming right along galvanized by our vision of the future. A vision encompassing three core tenants, the acceleration of the discovery of a viable cure, the development of improved treatments, and the enhancement of quality of life for those with cystinosis. I speak about these core values a great deal because they are the framework on which incredible ideas are built.

With regards to curative therapies we are ever-hopeful as the current five dosed patients still remain off oral cysteamine post gene therapy and study results show a continued favorable safety profile!

Leadiant Biosciences and CRN teamed up this past Spring for another Town Hall Meeting to discuss all things Cystaran (an immediate release eye drop to reduce corneal cystine crystals) and its re-introduction to the public. The back-of-house work and countless hours spent to get this medication back on the market was incredible.

CRN continues to give excellent support to families through a litany of programs, initiatives, scholarships and grants, all of which could not be possible without the help of an amazing array of volunteers and staff who juggle life's ups and downs; most doing so with the backdrop of either caring for a loved one with cystinosis, or are a patient themselves. It's an incredible bunch of humans all working towards the same goal: make cystinosis a disease of the past, while navigating the best individualized route to achieve that.



Jon and Elle

No cystinosis patient is exactly the same, we are a unique and rare mosaic of presentations hailing from all reaches of this world, speaking different languages, from all walks of life, requiring nuanced and patient-driven care. Through the CRN, new families/patients initially find commonality in our disease, but soon find the side benefits of fellowship, real-time answers from real patients/caregivers/clinicians, and a literal "network" encompassing an incredible array of services ready to be discovered.

It is our foundational principle to be that beacon of hope in the dark moments of this disease. Cystinosis makes you feel like you are in the middle of a hurricane with only a small sailboat. No lights, no idea where the shore is, with no direction or compass to point towards safety. The CRN was just what we needed; a lighthouse in our family's greatest time of struggle who's bulb burned so bright, offering direction and a beam of hope when we thought there was none. Never, ever lose hope. For now and always, we will continue to find new and innovative ways to support this rare community, strengthen our bonds, and fight for what these rare warriors need and deserve.

- Jonathan Dicks

Cystinosis Warrior Impact Program

Year five was a significant milestone for our awareness campaign and deserved an equally important global initiative. The Cystinosis Warrior Impact Program Campaign was launched with a goal of supporting, helping or finding a way to positively impact every single one of our 2,000 cystinosis warriors.

Support can cover many categories including but not limited to:

- Mental health and wellness
- Financial (scholarships)
- Medical (support programs)
- Education
- Research opportunities
- Networking (mentor program, connecting with resources to find a kidney donor, find a doctor, etc.)
- Translation services
- Advocacy and policy work and more

“ Never be afraid to ask for help, we are here to help in any way that we can, us warriors need to stick together. ”

- Jana H.



Getting help for yourself, your family, or other cystinosis warriors

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.

Introducing . . .

THE CYSTINOSIS
WARRIOR IMPACT
PROGRAM

Goal: to positively
impact every single
person living with
cystinosis

The Birth of a New Journal: Journal of Rare Diseases (JORD)



Rare disease research includes clinicians, patients/families, biologists, geneticists, genetic counselors, advocates, funders and educators spanning various disciplines and interests. Technical advances and innovations in sequencing revolutionized rare disease diagnosis. This is crucial to ending the long odyssey of patients/families by unraveling the cause of illness and the way forward towards effective therapy and hopefully a cure.

[Journal of Rare Diseases \(JORD\)](#) is published by Springer and is the official publication of The Egyptian Medical Association (EMA) given the growing interest and steadily increasing publications in the field of rare diseases.

JORD is an open access, peer-reviewed journal with clinical and basic research interests in rare and genetic diseases. JORD has an internationally diverse Editorial Board with expertise spanning various disciplines. The journal welcomes submissions in all fields of rare diseases and provides a forum for discussions on cutting edge perspectives in the field of rare diseases.

JORD view rare diseases as an important public health issue given their collective impact on individual patients/families, rare disease communities and healthcare systems. As such JORD

[Continued on Page 7](#)



Cystinosis Awareness Day



We recently celebrated the five year anniversary of Cystinosis Awareness Day. May 7 (5/7) was selected because it recognizes the most prevalent CTNS gene mutation, a 57-kb deletion. As the name implies, awareness day serves as a reminder to educate others about this rare disease.

Since its 2017 inception, Cystinosis Awareness Day has become recognized by advocacy organizations, industry partners, individuals and families inside and outside of the cystinosis world. Over a dozen countries participated in this year's events. Of note, the Mexican Organization of Cystinosis AC held a Family Day in Puerto Vallarta. The gathering attracted medical professionals from Columbia, Mexico and the U.S. Learn more on page 5.

In the weeks leading up to May 7th, community members hit the pavement; interviews were lined up and prepared for, articles written, and almost every social channel had unique cystinosis perspectives popping up in our news feeds. Thank you to everyone who took a moment to "share their rare".

Here is a sample of the awareness day coverage:

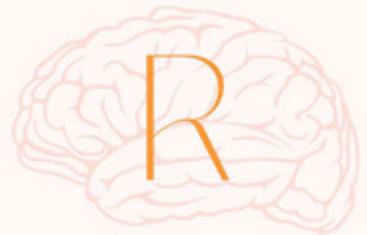


Dual-Perspective Interview with [Beacon](#) CEO



RARE MAMAS

Rare Mamas: A "Rare Bond" Connecting with Cystinosis
[Page 23](#)



IS FOR RARE

[R is for Rare Podcast](#) with Ashley Abedini



2022 Giveaway

As a thank you for growing with the CRN and our awareness efforts, plantable ribbons loaded with wildflower seeds are now available. Request one free of charge by visiting <https://bit.ly/3vDdHej>.

Cystinosis Family Day - Mexico 2022



Learning, fun and togetherness was the essence of the Cystinosis Family Day Mexico 2022, after a couple of difficult years, it was the opportunity to meet again to share and enjoy with Mexican families and newly diagnosed patients.

We enjoyed speakers of international stature who informed us about the current situation of cystinosis, as well as we decided to address important issues that are useful for patients and their medical management.

Physicians from Colombia, Mexico and USA virtually participated, highlighting the following topics:

- General overview on cystinosis and importance of treatment of eye drops and the way it acts in corneas
- Patients' rights and commitment of hospital institutions in relation to rare diseases in Mexico
- Recommendations on care in Fanconi Syndrome, generalities of cystinosis and medical advances in transplants and gene therapy
- Current overview of Rare Diseases in Mexico, statistical data, and the importance of organizations that work on behalf of patients with these low prevalence conditions
- Mental health care for patients and their families

It is important to mention that many of the aforementioned topics discussed at our annual meeting are specifically selected for Mexican patients, due to the country's current health and governmental situation.

The Mexican Organization of Cystinosis AC in recent years has given support to patients and highlighted the issue of mental health, has intervened so that everyone has their comprehensive treatment for cystinosis given by government health services, and has collaborated with doctors from various specialties to give the patient the proper medical care for this condition.



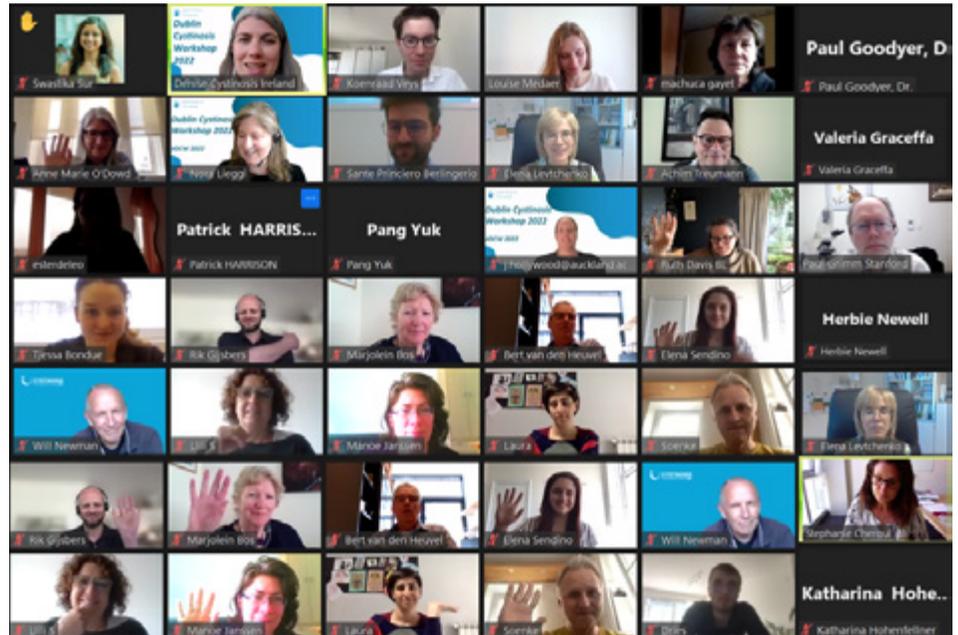
Dublin Cystinosis Workshop

By Denise Dunne

The annual Dublin Cystinosis Workshop (DCW) was held online on Thursday 21st April 2022, creating collaborations and conversations between world class researchers and clinicians. Now in its eighth year, this unique event allows those active or interested in cystinosis research to share their insights and most recent findings as efforts continue to identify new drug targets and effective therapies for cystinosis and ultimately to find a cure for this ultra-rare disease.

Several new treatments were discussed at the meeting; for example, a proof-of-concept study found that the gene MFSD12 may be a viable therapeutic target for a new class of cystinosis drugs. Meanwhile, a drug combination of cysteamine and everolimus has shown promise in mice – addressing issues caused by cystinosis while also preserving kidney function. Cell and gene-based therapies are showing curative potential and research in this area is making significant ground. According to studies carried out in mice, early therapy with a drug called ELX-03 could prevent progressive Fanconi Syndrome. Excitingly, the prodrug CF10 is due to start clinical trials in 2024. In addition, new therapies may be en route for issues such as cystinosis myopathy.

Efforts to improve the quality of life of cystinosis patients are also bearing fruit; for example, it was reported that whole body vibration therapy is safe and effective and early results show that an exercise programme that incorporates it appears to help improve muscle strength and balance in people with cystinosis. One of the keynote scientific speakers, Dr



Andreas Natsch, head of in vitro Molecular Screening at Givaudan Schweiz in Switzerland, outlined how body odour has a significant impact on quality of life for cystinosis patients. He explained that scientists had recently gained a better understanding of the processes involved in odour release, and thus may be able to manipulate the release of the odour-causing compounds. New vitamin E technology used in contact lenses appears to allow for the sustained release of cysteamine for as long as eight hours, meaning administration of large quantities of eyedrops could be a thing of the past for cystinosis patients.

And as cystinosis patients live longer, healthier lives, issues such as fertility in cystinosis patients are finally being addressed. The molecular basis for fertility issues are being examined and are close to being understood and this may open up new therapeutic avenues.

Another keynote scientific

speaker, Professor Paul Grimm, Professor of Paediatrics (Nephrology) at Stanford University, outlined a groundbreaking new procedure whereby a combined stem cell and kidney transplant are carried out at the same time with a view to leaving the patient drug free.

A fitting presentation was the one given by Public Patient Involvement Manager at the RCSI University of Medical and Health Sciences in Dublin, Ms Lorna Kerin. She noted that patient involvement in research from the very earliest stages is not just a “nice to have” but a must-have. This involvement must be meaningful and not tokenistic. Cystinosis Ireland is well placed to lead high impact PPI, with a well-motivated and highly educated patient population. “People with cystinosis have the expertise on how to live with the disease but also they have educated themselves beyond what people living with other more common diseases have done,” agreed Ann Marie O’Dowd of Cystinosis Ireland.

The winner of the 2022 Prof Roz Anderson Memorial Prize was Elena Sendino Garv. Elena is a PhD candidate at the Utrecht Institute for Pharmaceutical Sciences (UIPS), in Utrecht, the Netherlands. The Prize is awarded to the best short science communication at the Dublin Cystinosis Workshop, as voted on by the attendees and celebrates the life and work of Prof Anderson, a staunch supporter of the cystinosis community and a leading researcher in the field. Elena’s current research focuses on kidney ciliopathies and tubulopathies, with the aim of using drug repurposing approaches and gene-editing (Crispr-Cas9) to restore the cell function of these kidney conditions. At the moment, she is working on the development of a

new 3-D bioengineered model for nephropathic cystinosis, in addition to the restoration the gene function and phenotype of cystinotic cell lines using Crispr-Cas9 HITI approach.

The Workshop also took the opportunity to celebrate the tremendous work by Dr Don Cairns and Dr Bill Gahl and the contributions they have made to the cystinosis research and patient communities in their careers. We also took the

opportunity to pay tribute to the late Dr Jerry Schneider who is sadly missed.

The DCW creates a creates a sense of community among researchers, clinicians, family members and those living with cystinosis. It creates a sense of belonging to something special with a shared common goal. We hope our 9th annual DCW 2023 will take place in person and will be just as impactful as this year’s event.

“ People with cystinosis have the expertise on how to live with the disease but also they have educated themselves beyond what people living with other more common diseases have done. ”

The Birth of a New Journal: Journal of Rare Diseases (JORD), continued

provides a dedicated platform for rare diseases research infrastructure to share knowledge and experience, to bring diagnoses and therapeutics to patients with the ultimate goal of improving quality of life.

JORD also welcomes submissions not only within single rare disease, but also between rare diseases. Articles with an integrated approach to coordinate efforts and potentially speed the development and the implementation of novel therapeutics aiming at cure not just effective treatment are encouraged.

Another area of rare disease promotion and research is equity. Articles addressing the challenges to bridge the healthcare gap across rare diseases and highlighting equitable benefits of research outcomes in different populations or socioeconomic groups are of paramount importance and highly welcomed at JORD.

JORD online submission is now open and authors are provided with editorial support throughout the writing and peer review process. For submission guidelines, [click here](#).

Prof. Neveen A. Soliman (pictured)
JORD Editor-in-Chief



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We're offering a **FREE one month's supply** (4 bottles) of CYSTARAN to eligible patients with a voucher*



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*Limit one offer per patient. No purchase is necessary. Offer valid in the United States and Puerto Rico, and void where prohibited by law. Not valid in the Commonwealth of Massachusetts. Subject to eligibility and terms and conditions, which are subject to change. Offer expires December 31, 2022.

Thank you for your continued loyalty and support.

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Education and Awareness Committee Update

By Marybeth Kruppenacker, Vice President of Education and Awareness



As we enter the second half of 2022, the world of rare diseases is always changing. It is hard to believe we have passed the second year of COVID and the changes that we have all had to make in our lives everyday. It is a new world.

As I reflect, I think about how fortunate the cystinosis community is. I certainly don't feel lucky to have to deal with and live with a rare disease everyday but I believe that we are lucky to live in a time of great discovery.

Advocacy organizations like CRN are on a cutting edge of discovery with new and improved treatments. Over many years we have developed and continue to develop relationships and partners in the pharmaceutical world to communicate with and work together with to aide in new treatments.

The third CNE (Cystinosis Network Europe) International Cystinosis Conference took place in Belgium July 7-10. Patients and doctors from around the world met in person to present and discuss the latest findings related to the world of cystinosis research. CRN was proud to participate and support the agenda throughout the sessions.

Cystinosis Research Network is once again exhibiting at the American Society of Nephrology meeting this fall in Orlando. For the first time in over two years we will be able to meet in person and network with industry and academia and share the latest in kidney transplantation and the world of nephrology. It has always been one of CRN's missions to be at meetings like this to educate the doctors about cystinosis. The goal at these meetings is to share information about the latest in cystinosis research and to advocate and educate for the community in general. It will be great to meet face-to-face once again!

Finally, I think one of the most important community events within the last year is having Leadiant Pharmaceuticals' CYSTARAN eyedrops back to market. Due to an unforeseen manufacturing issue, production of these critically important eyedrops was stopped. Leadiant remained committed to our



small, rare disease community and through a long, arduous process of FDA regulations and trial, the eye drops are once again available to our community. While it was a very difficult time for many of our patients who were not able to switch to CYSTADROPS, Leadiant was a true partner and kept the CRN informed throughout. As I have said many times before, the CRN community is blessed to have options with our medications; we have choices because of the advocacy and partnerships and CRN remains strong in that commitment!

Since our last newsletter, we had two former CRN Board members pass away. I reflected briefly on both of their incredible lives (please continue on to page 9). We are thinking of their families, friends, and the many lives these women have impacted.

In Memoriam

Cheri Friend

By Marybeth Kruppenacker - February 19, 2022



I have been thinking a lot about my friend... CheriFriend (I never just called her by her first name) this week since she passed away Tuesday night. I have read all of the wonderful things her family has said about her and I have come to realize even more that she was just a joyful and loving person. Her life was about her family and friends and they loved her so. She certainly was someone of great faith and I think she demonstrated that as she fought cancer with faith, strength and grace. Her willingness to

help everyone was always evident... it was never about CheriFriend... it was always about you! She was one of the first people to call me after Laura's [cancer] diagnosis to offer me encouragement and prayers. I will never forget that! So today Laura will be with your beautiful family celebrating your life. I know there will be tears and most certainly laughs, but most importantly there will be love and memories for YOU my dear friend... CheriFriend... thank YOU for being my friend and "cysta."

Jessica Britt Jondle



It is with a great sadness that we announce the passing of one of our former Board members, Jessica Britt Jondle.

Jessica was 41 years old, a wife, mother, writer, and educator who wanted to make life better for everyone living with cystinosis. She was one of the earliest advocates for adults with cystinosis who, she felt, were not always considered in the world of research and support. Whether you agreed or disagreed, Jessica wanted what was best for all cystinosis patients.

Despite a cystinosis diagnosis she lived a full life and with her perseverance and fight; she even took up hiking when people didn't think she could or should...but she did!

Jessica hiked the John Muir trail, and summited many mountains with the highlight being her summit of Mount Kilimanjaro.

Jessica took part in research protocols when given the opportunity and was committed to making life better by participating when asked. She was a strong, independent woman who wanted to be seen as just that - not defined by her diagnosis. She never wanted to be told something was unattainable and she certainly did not let this illness stop her from living her best life.

Rest in peace, Jessica, and thank you for doing your part to make life a little better for all. Our deepest sympathy to her husband Wayne and two young children.

Remembering Dr. Jerry Schneider

Dr. Schneider focused on the study of metabolic disease for over 45 years. While at the National Institutes of Health (NIH) he began researching cystinosis - research that would lead to the creation of cysteamine treatments many take today. For the countless lives he touched, we wish you all comfort during this time. He was a true pioneer and consummate advocate in the fight against cystinosis.

Obituary and photo published by La Jolla Light/legacy.com.

Dr. Jerry Allan Schneider, Professor of Pediatrics at UCSD School of Medicine from 1970-2011, died December 28 at his home in La Jolla, California. The cause of death was respiratory complications of Parkinson's Disease.

The eldest of four brothers, Dr. Schneider was born in Detroit, Michigan in 1937. His father Benjamin emigrated from Russia at age 13, and eventually owned and operated a dry goods store in Detroit. His mother Sarah (Sally) was a homemaker.

Dr. Schneider graduated from Cass Tech High School, and then attended the University of Michigan. He received his medical degree from Northwestern University, followed by a pediatrics residency at Johns Hopkins University and a fellowship in inherited disorders of metabolism in the laboratory of Jay Seegmiller at the National Institutes of Health.

During his internship in Baltimore, Dr. Schneider met and married Elaine Bergner of New York, then an undergraduate student at Goucher College.

After the NIH, Dr. Schneider spent his career as a professor at UCSD School of Medicine, where he was one of the

founding members of the Department of Pediatrics. For the last eight years of his career, in addition to his research and patient care activities, Dr. Schneider also served as Dean for Academic Affairs.

Dr. Schneider spent his entire career working on a rare genetic disease, cystinosis, a lysosomal storage disorder. The disease is caused by a gene defect which prevents the amino acid cysteine from being transported out of lysosomes (the cell organelle in which proteins are broken down to their component amino acids). Without treatment, cysteine crystals accumulate intracellularly, which leads most notably to kidney failure. Over the course of his career, Dr. Schneider and teams of colleagues sorted out the inheritance pattern of the disease, the nature of the defect, and eventually came up with a medication to treat the disorder. Prior to this research, children with cystinosis died at an early age. In the current day, patient survival is excellent, and some of the early patients have even gone on to have children of their own.

In addition to his scientific research, Dr. Schneider was extremely devoted to his wife of 58 years and their two daughters, Danielle and Jane. As a father, he emphasized the importance of hard work and education, and was known for retelling a stable of stories that demonstrated the importance of doing the right thing. These were jokingly referred to by his family as "Dad's morality tales".

Dr. Schneider's other life passion was saltwater fishing. He made his own rods, filleted his own fish, kept a dry-smoker in the garage, and experimented with pickling and canning. He developed a supportive



community of fishers from among his colleagues and neighbors, frequently chartering several-day trips off the coast of Southern California and Mexico. Ever the scientist, he kept a log-book of his fish recipes, recording differing ratios of each ingredient, and noting the results. The final pages of this log-book simply read "excellent, excellent, excellent". In keeping with his sense of community, colleagues, friends and neighbors were regularly invited over to pick up gifts of freshly caught, pickled, canned and smoked fish.

Dr. Schneider is survived by his wife Elaine, daughters Danielle and Jane, son-in-law Keith Eaton, three granddaughters, Rachel, Aliza and Nina, and his brother Rabbi Paul Schneider of Baltimore. He was pre-deceased by two brothers, Mark and Larry.

Family Support Committee Updates

By Jen Wyman, Vice President of Family Support

“Your support network is the solid ground from which you can propel yourself upwards.”

- Anna Barnes

Having social support networks is an important and necessary part of navigating our lives with cystinosis. Whether we are patients, caregivers, spouses, siblings, grandparents, etc. we all need a strong support system. Hopefully, you have all found some of that within the Cystinosis Research Network and its members. We are a small community, but a mighty one and with the help of social media we are able to get connected and stay connected. But we also need informational support. Navigating the world of rare disease leads us to unfamiliar places; insurance, medical, education, daily life, mental wellness... the list goes

on. The Cystinosis Research Network is continually trying to find ways to fill those needs. It is a constantly evolving task and an important one. Our website is an amazing resource for support, whether it be family, medical, insurance or otherwise. There are programs like the Live Like Laura Fun Fund, which honors Laura McGinnis and empowers cystinosis warriors to create their own memories; The Cystinosis Memorial Fund, which honors those who have lost their battle with cystinosis, and provides financing for technology and career support to teens and adults with cystinosis; and the Care Package Program which provides “pick me ups”

to those who are struggling. These are just a few of the support resources CRN offers. And most importantly we have people... people who have been where you have been, or are where you are, and we can connect you to help you navigate your journey with cystinosis in whatever capacity needed. So, reach out... reach out if you need someone or something. We have big-hearted, giving, knowledgeable humans of all ages, from all around the world, who can and want to help. You don't ever have to feel like you are alone.

Reach out to one of us anytime:

✉ jenwyman@comcast.net
Vice President Family Support

✉ Kristinasevel@gmail.com
Board Member; Family Support

🖱 Programs mentioned can be found at cystinosis.org/special-programming



Care Package Program

 CYSTINOSIS RESEARCH NETWORK

“What a really nice idea to cheer those who are down up. The kit was such a kind gesture and I'm certain it will bring cheer to anyone in need.”

-Joan





Living with Cystinosis Support Group

A supportive, web-based
peer support group for those
living with cystinosis
facilitated by

Kerry Heckman, MSW, LICSW

3rd Tuesday of each month from
4-5pm PST / 7-8pm EST

To sign up, visit
thecenterforchronicillness.org/groups



Contact us at
info@thecenterforchronicillness.org
or (425) 296-2705 with questions.

www.thecenterforchronicillness.org

This program is free of cost.



Center *for*
Chronic Illness

Supporting Loved Ones of those Living with Cystinosis Support Group

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

3rd Tuesday of every other month
(March 15th, May 17th, July 19th, Sept 20th, Nov 15th 2022)
6-7pm (PST) / 9-10pm (EST)
To sign up, visit www.thecenterforchronicillness.org/groups

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



Life Beyond Cystinosis

By Shannon Keizer

If you've spent time with the cystinosis community, you've probably heard the phrase "Cystinosis doesn't define me". And it's true. Each of us is so much more than our disease. We are working professionals. We are students. We are parents, spouses, aunts, uncles, siblings, and friends. We are active members of our local communities, bloggers, adventure seekers, and we are trying to make our mark on the world however we can. Cystinosis does not define us.

Then, there are the moments we can't deny it. Those unfortunate times when we are faced with the reality of our disease. I'm sure we all have our hospital speeches memorized. Yours might sound something like, "Shannon Keizer. April 16, 1989. I'm here because I have left sided flank pain, nausea, increased heart rate, and a fever. I have a rare, genetic disorder called cystinosis. My GFR is around 24. I have a history of kidney stones, c-diff, and sepsis". The interview continues, and everyone wants to know more about this rare disease you have.

We have become experts in our own healthcare needs. We know our bodies, and we can usually tell what's wrong. We know the healthcare system. We can tell the difference between the caregivers that live up to their title and those that don't. We know which ones truly listen and which ones empathize with our stories.

In fact, it was an apathetic nurse that made one of the biggest impacts in my life. For almost 20 years, I dreamed of becoming a nurse, but my dislike of science and fear of the classes stood in the way. Microbiology



and biochemistry were never my forte. The unpleasant encounter with this nurse and catching a near med error gave me the confidence and motivation to pursue my dream. I wanted to be a "good" nurse.

My first semester of pre-reqs was exactly what I expected. HARD. Day one of physiology and the professor was already talking about active and passive transports and diffusion of molecules. No freebie syllabus day for this nursing program. The next three years would follow suit. I studied, studied, and studied more. Almost daily from 9am to 10pm. I lived and breathed nursing and science classes. When I wasn't studying, I was working a part time job, serving at church, and tried to carve out a few hours per week for something fun. I often wondered whether the intense schedule was worth it. I excelled in my classes, but it was mentally and physically draining.

About a year into the program, my very first clinical day arrived: 4:30am wakeup for a 12 hour shift. To say I was nervous is an understatement. But the moment I walked through the hospital doors, donning black scrubs and a stethoscope around my neck, the nerves turned to overwhelming emotion. For 30 years, I was the patient in the hospital bed. Out in the world I was Shannon, but in here, I was the girl with cystinosis. Today, for the first time in a hospital setting, I wasn't the girl with the rare disease. I was a professional on the other side of the bed. Tears filled my eyes.

Patient experience prepared me for all the things school can't teach- empathy, advocacy, active and attentive listening, patient education, treating people as more than a diagnosis, and little things like extra ice in water cups and stirring MiraLAX until it is completely dissolved. Being a patient did not prepare me for things

“ Today, for the first time in a hospital setting, I wasn't the girl with the rare disease. I was a professional on the other side of the bed. Tears filled my eyes. ”

like drainage tubes, ostomies, trach care, NG tubes, TPN feedings, PICC lines, care plans, mechanism of action for each drug given, head to toe physical assessments, neurovascular assessments, IV piggybacks, and so much more. I can't count the number of times I felt ill-equipped to be a nurse. I often questioned if I had what it takes.

During those moments, I reminded myself of all the reasons I pursued nursing in the first place. I reflected on the personal connections I'd already made during my short time in scrubs. I recalled the non-verbal, elderly woman with dementia, c-diff, and sepsis who needed an advocate that understood what she was going through. The teenage girl with who had been in and out of the hospital multiple times with severe depression. And parents of the little boy with failure to thrive and a newly placed g-tube who needed help scheduling doctor visits.

I realized that people didn't expect me

to have all the answers. They didn't mind that I was clumsy or hadn't yet mastered the skills. They just wanted someone to see them as humans, listen, and be their advocate. I was reminded of the phrase, "If God called you TO it, He will call you THROUGH it".

Many people came along side to encourage me. Their support helped carry me through the most challenging moments. Nurse friends insisted nursing school is supposed to be hard and that I certainly had what it takes. Hearing those words from people that have been in the same position gave me the confidence to say, "You're right. I can do this". And indeed I did. I graduated this past May with my RN degree!

I currently know of a few within the cystinosis community that are pursuing nursing degrees. Many more are chasing other big dreams, and some have been contemplating taking the leap to begin something new. If this is you, I want to give you the



same encouragement many others gave me. If you have a dream, do it. If something is hard, do it anyway. Often the hardest things are the most rewarding. Go at your own pace. Remember your "why" and keep that in perspective. You are more than your disease, and you ARE making a difference in the world!

Thank you to all those within the cystinosis community that helped along my journey. Your words brought me through some difficult times, and I hope I can do the same for others.

Live Like Laura Fun Fund Update



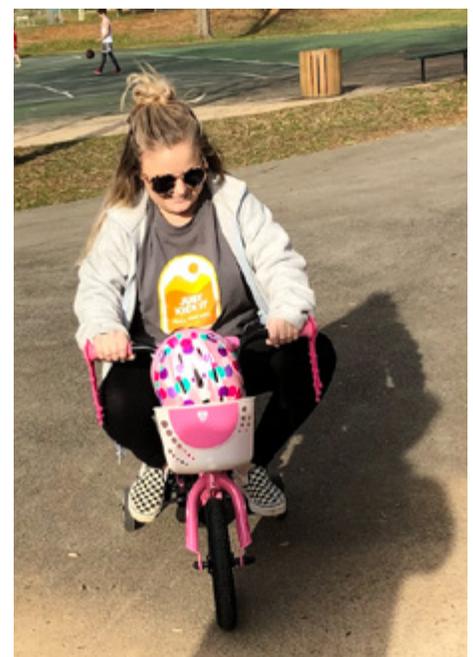
The vision of the Live Like Laura Fun Fund (LLLFF) is to enable people affected by cystinosis to participate in life's many adventures.

Our "squirrel", Laura McGinnis, was a woman who lived life largely and loudly. The idea that she could not do something (cystinosis or not) was foreign to her- she found a way. In her short time with us, Laura was

determined to make the moments count.

Through the LLLFF, we hope to empower cystinosis warriors to create their own memories. Need help buying that concert ticket? Purchasing a theme park pass to ride the largest rollercoaster? Trying to conquer that rock climbing wall? Whatever it is that may bring you joy, we want to help you seize the day!

People affected by cystinosis and their siblings may apply for up to \$1,000 USD. For questions, email Frankie at frankiemcginnis@gmail.com or get started at cystinosis.org/lllff.



You are never too big or old to claim adventure - even on a baby bike.

Rare Disease Day 2022



Observed on the rarest day of the year, February 28th marked the 12th annual Rare Disease Day. The global event campaigns for equity for the over 7,000 rare diseases currently identified. Cystinosis resides among those rare diseases; known to have a population of 2,000 worldwide with 600 residing in the U.S.

This year, the CRN and RareDiseaseDay.org shared ways to make a better world for those living rare. Virtual events included Eurordis' Global Priority for Equity Summit, EveryLife Foundation's Rare Disease Week on Capitol Hill, Rare Disease Week with the National Institutes of Health, patient panels with Wego Health, the Rare Disease Diversity Coalition and Black Women's Health initiative, and more. CRN President Jonathan Dicks appeared with daughter Elle on WTNH Good Morning Connecticut and as a guest speaker for Quinnipiac's Rare Disease Day Symposium. In addition, Steve Schleuder and Christy Greeley recorded testimonials as part of the Rare Disease Day Bay Area's Rare Disease Week festivities.

Toolkits for Tough Conversations

School Toolkit. The kit may help you have discussions with teachers or young children about rare disease.

Equity Toolkit. Featuring tools to assist people living with a rare disease improve social opportunity, non-discrimination in education and work, and equitable access to health, social care, diagnosis and treatment.

Resources can be found at rarediseaseday.org and cystinosis.org.

953 RARE DISEASE ADVOCATES
370 MEETINGS WITH CONGRESS
297 PATIENT ORGANIZATIONS
49 STATES + D.C, PUERTO RICO AND VIRGIN ISLANDS
1 AMAZING RARE DISEASE WEEK!



rareadvocates.org/rdw

#raredc2022

Development Committee Update

By Jonathan Dicks, Vice President of Development

As we are in full summer swing with sports, managing gardens, getting kids off to summer camps, and family vacations it's hard to find any downtime, good thing we are innate multi-taskers I guess. Here on the development-side of things "downtime" is non-existent as well and we like it that way truthfully. That means we're hard at work not only cultivating the existing relationships with our incredible industry partners, but also traversing other non-traditional advocacy avenues outside of our rare community. This year we have secured over \$275,000 in grants, not including community-driven fundraising initiatives! I find myself yet again humbled by the generosity of our industry partners, Horizon Therapeutics, Leadiant Bioscience, Recordati Rare Disease and AVROBIO who all share the same conviction that every person with a rare disease has the right to the best possible treatment.

This May 7th saw our 5th Annual Cystinosis Awareness Day in full bloom with incredible community-led advocacy efforts. In honor of our 5th year celebrating these incredible warriors we call family and friends, we had a lofty idea...here me out. The question was posed, "if we really are the CRN, then what can we do to increase the 'network' portion of who we are?" A "network" is synonymous with a framework, and has connotations of facilitation. Out of this question was born the CWIP (Cystinosis Warrior Impact Program) with the singular goal of positively impacting every person living with cystinosis. We hope to accomplish this by utilizing our vast

resources, examples could include: matching a newly diagnosed caregiver with an experienced family, helping prepare a cystinosis teen with tutoring assistance, providing an adult with mentor services or training them to give back by becoming a mentor, pairing families with physicians experienced with cystinosis, or providing mental health resources/support groups/access to the Adult Leadership Advisory Board members. This is a long-term goal and will only be possible with the assistance of our network of patients, caregivers, loved ones, partners, and healthcare professionals. We hope you feel empowered to take part in this endeavor to positively impact our rare disease community. Interested? You can email info@cystinosis.org to get started AND to refer a friend for services.

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

Bromberg & Associates who offered the CRN translation and telephonic interpretation for Spanish-speaking individuals and families at our 2021 Virtual Family Conference are back and have expanded their reach to cystinosis patients all over the world offering translation services in over



The Dicks Family

200 distinct languages, with an ever-expanding list. See page 15.

CRN and the Center for Chronic Illness are offering free, professionally-facilitated support groups and health education programs for those impacted by cystinosis as well as a separate monthly group specifically for parents with a diagnosed child. See pages 17 and 18.

The Avocet Course at Wild Wing Plantation in Myrtle Beach hosted Gail Potts' 2nd Annual Cystinosis Research Network Charity Golf Tournament on June 18th. Incredible advocacy work Gail, we are so appreciative of your dedication and grace. We can't wait to watch this event grow next year!

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



Now over 200 languages available!



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TRANSLATION AND INTERPRETING SERVICES

Cultural Awareness Programs • Interpreting and Translation • Video Remote Interpreting
Compliance Consulting • Website Localization • Language Training • Relocation Services

**Bromberg's Translation and Telephonic Interpreting Services Program,
sponsored by Horizon Therapeutics**

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

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

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

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

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

OPTION 1: Connect with an interpreter by telephone:

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

OPTION 2: To obtain a document translation:

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

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

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

C-HZN-00329



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Phone: 313-871-0080

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Cultural Awareness Programs • Interpreting and Translation • Video Remote Interpreting
Compliance Consulting • Website Localization • Language Training • Relocation Services

Programa de servicios de interpretación telefónica y de traducción de Bromberg patrocinado por Horizon Therapeutics

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

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

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

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

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

OPCIÓN 1: Contactarse con un intérprete por teléfono:

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

OPCIÓN 2: Para obtener una traducción de un documento:

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

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

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



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Honoring Her Memory

By Gail Potts

My daughter Deanna Lynn was diagnosed with cystinosis when she was 10 months old. Definitely not the norm. We were fortunate enough to be living in Houston, TX at the time and had some astute physicians. Her symptoms were those of most of the children with vomiting and excessive thirst and volumes of urine excretion. She also developed a yeast infection which would be strange for an infant. We did a dipstick urine test which showed high sugar and protein in her urine, making a tentative diagnosis of diabetes and immediate hospitalization. Of course this led to consultations with metabolic disease specialists because blood glucose was normal, but electrolytes were all off. Her diagnosis was made by doing conjunctival biopsies and bone marrow aspiration. This took place 49 years ago. What remarkable advances have occurred.

Although there were too many hospitalizations to count over Lynn's 27 years, she managed to enjoy Girl Scouts, dancing, and drama in high school. She attended her Junior and Senior Proms which were a couple of the truly exciting times in her life. She was discharged from the hospital so she could go to the prom on the condition she would be readmitted the day after.

Lynn had her first kidney transplant when she was almost nine years old and her second when she was 17 years old. Both being cadaver transplants. She didn't start on oral Cysteamine and eye drops (necessary cystinosis treatments) until she was about 16 years old. Her initial visit to the National Institutes of Health (NIH) was when she was 13 years old. At that time they were not offering

Cysteamine to transplant recipients. However, when her eyes were so painful from the crystals we embarked on another NIH visit and she was included in the studies. Although NIH was not sure whether growth hormone would be beneficial, she began taking it when she was about 20 years old. She did benefit some from it.

Lynn graduated from a junior college, but found a baccalaureate program more challenging and had to drop out. After exiting college, she studied to become a medical assistant and worked part time in a clinic.

This is the cliff notes of her journey as this could become a novel and everyone with cystinosis has been down this path. Lynn loved children and enjoyed babysitting. She had a heart of gold. She was always generous with what she had. Employment was difficult because "she looked different". Accommodations for her size and health issues which employers didn't want to address, I think, contributed to her inability to get work.

To honor her memory and generosity we started the Deanna Lynn Potts Scholarship for individuals with cystinosis 22 years ago. I am happy to say we have awarded 17 academic scholarships. Now I'm honoring her by joining the Board of the Cystinosis Research Network. This position has led me to the challenge of fundraising; not my strong suit, by any means. Last year was my first attempt at a fundraiser; a golf tournament in Myrtle Beach, SC. June 18, 2022 was my second effort at the Wild Wing Advocate Golf Course. Although the event is over, the fundraiser will remain open if you'd like to consider



Deanna Lynn Potts

making a donation at cystinosis.org/howtohelp-donate.

Multiple scholarship opportunities, including Deanna's, now accepting applications at cystinosis.org/support-resources/scholarships.

Apply Now!

#cystinosisaware



ACCEPTING
APPLICATIONS
FOR 2022
SCHOLARSHIPS

academic &
vocational

The efficacy you can see. The convenience that matters.

- Cystadrops dissolves cystine crystals throughout the entire cornea, not just the top layers
- Cystadrops showed a 40% decrease of cystine crystals in the cornea after 90 days of treatment
- Cystadrops has convenient 4 times a day dosing

 **Cystadrops**[®]
(cysteamine ophthalmic solution) 0.37%

Actor Portrayal

Indications and Usage*

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

Important Safety Information

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

* For use by individuals with cystinosis

Visit www.cystadrops.com for full prescribing information



 **Cystadrops**[®]
(cysteamine ophthalmic solution) 0.37%
Special drops for special eyes...your eyes

A “Rare Bond”: Connecting with Cystinosis

By Shirley Dicks and Tori Westfall



SHIRLEY & ELEANOR

After two frustrating years of tirelessly working with our GI team at our local pediatric hospital, we had zero medical explanations of why our three-year-old daughter Eleanor (Elle) was constantly throwing up. She had no appetite, was excessively winded, and had an aversion to sunlight and bright lights. Our daughter was not growing, so we chose to see an endocrinologist who specializes in pediatric growth disorders. After reviewing her labs and discussing Elle’s presenting symptoms, he had an unofficial diagnosis. On October 10th, 2018, Eleanor was diagnosed with cystinosis. Being without treatment and therapies for so long had left Eleanor in stage 3 kidney failure. All of this news came as I was 20 weeks pregnant with our second child. We were then met with the realization that this disease was genetic, with a 25% chance of our unborn child presenting just like Elle did. The waking hours of life at home were filled to the brim with appointments and around-the-clock medications. Things were hard, and

I was exhausted. Family and friends were there to provide hugs, meals, and encouragement, but no one truly “got” what raising a child with a rare disease actually entailed from one uncertain day to the next.

Life after receiving a rare diagnosis felt like trying to keep our heads above an ever-rising current. Some days felt like smooth sailing, while on other days, we barely took a breath. It takes time to get into the “new normal.” I am grateful for finding our second family in the Cystinosis Research Network. There is a community of rare families and other moms like me who are going through similar experiences and can intimately relate.

We are year three post-diagnosis, and I can say we finally have our heads out of the muddy water for now. I’m forever grateful for the rare mamas that helped me get through those days when it felt like we were just sinking; without them, we might still be stuck. I was able to connect with Tori, whose daughter has cystinosis after she posted about their struggles with medications and the same cyclical

vomiting that Elle had once suffered from. She was up to her ears in the muck, right where we were three years ago, and we knew we could offer a rescue line to help pull her out.

TORI & ADDYSON

My three-year-old daughter, Addyson, was diagnosed with cystinosis, Fanconi Syndrome, and Kidney Disease on May 10th, 2021. Since birth, I was concerned about Addyson’s acid reflux and constant spitting up, but after the age of one, she stopped growing and gaining weight. Her pediatrician at the time showed no concern.

After three months of no improvement or weight gain, I switched treatment teams. Addyson had no improvement after three more months, but the team saw no other sign of concern. They referred us to an Endocrinologist. From there, we were referred to Nephrology. Thankfully the Nephrologist was familiar with Fanconi Syndrome and cystinosis, recognizing it immediately from the initial blood work. Addyson was hospitalized and had a g-tube placed for the

administration of her many medicines and nutrition. A few days following, the WBC level results confirmed cystinosis. Since then, Addyson has been admitted to the hospital three separate times this year and has had over 20 doctor appointments.

There are times other cystinosis mamas know what is going on with my child before the doctors do. Shirley introduced me to the idea of using a combined GJ-tube over her current G-tube only. She also expressed that Addyson's symptoms closely mimicked Elle's battle with Gastroparesis (Delayed Gastric Emptying). I shared my concerns

with our treatment team, revealing a positive diagnosis of Gastroparesis for which she is now being treated. A second rare mom helped facilitate connection with a specialty compounding pharmacy service, which has saved me countless hours of drive time monthly. The support, caring, and understanding are unmatched in the cystinosis community.

It is so important for rare mothers to connect with others for knowledge, encouragement, and support through such unknown and uncommon circumstances. Even the doctors familiar with cystinosis struggle

because each patient presents with such a variance in symptoms and barriers. When you have a child with a rare disease, it is very lonely and isolating at times because your friends, family, and peers will never fully understand. What I have come to realize is that, in fact, there are many mothers who have been where I am, ready to help at a moment's notice; I just had to reach out.

No matter what path we go down with this disease, we are better together than apart.

Originally published as a guest article on raremmas.com.

The graphic features a woman in a blue shirt and dark pants sitting on a rocky shore, looking out at a large body of water under a cloudy sky. The image is overlaid with two large blue circular shapes. The left circle contains a quote in white text: *"When I was awarded with the Cystinosis Memorial Fund (CMF), I was given the gift of a peace of mind. I am currently beginning a masters program in counseling psychology. I look forward to becoming a therapist one day and looking back on the peace of mind I experienced in the beginning thanks to the CMF."* Below the quote, the name **CHERYL S.** is written in white. To the right of the name is a circular logo consisting of several green human figures holding hands in a circle. The right circle contains the text **Cystinosis Memorial Fund Recipient** in white.

Information and online application now available at cystinosis.org/cmfi.

HAVE YOU ATTENDED AN IMPACT PROGRAM YET?

The IMPACT Program is specifically designed for people living with cystinosis and their families, and sponsored by Horizon Therapeutics throughout 2022



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

Even if you've attended an IMPACT Program previously, please check back in with us!

The IMPACT Program has new topics created specifically for different cystinosis patient experiences—**adults, teens, and families/caregivers** of people living with cystinosis.

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

USE AND IMPORTANT SAFETY INFORMATION

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

PROCYSBI can cause serious side effects, including:

- **Skin, bone, and joint problems.** People treated with high doses of cysteamine bitartrate may develop abnormal changes of their skin and bones, such as stretch marks, bone injuries (such as fractures), bone deformities, and joint problems. Check your skin while taking PROCYSBI. Tell your doctor if you notice any skin changes or problems with your bones or joints. Your doctor will check you for these problems.
- **Skin rash.** Skin rash is common with cysteamine bitartrate and may sometimes be severe. **Tell your doctor right away if you get a skin rash.** Your dose of PROCYSBI may need to be decreased until the rash goes away. If the rash is severe, your doctor may tell you to stop taking PROCYSBI.

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





The Adult Leadership Advisory Board (ALAB) is a group comprised entirely of adults living with cystinosis.

ALAB's mission: To share our stories and strength to educate, motivate and empower the entire cystinosis community. Through partnerships with the CRN and other organizations, ALAB focuses on issues and challenges through developing programs, opportunities, and mentorship. Our goal is to create a group that will not only benefit from the experience but will contribute to the entire cystinosis community.

Now Accepting New Member Applicants

If you are interested in joining ALAB to help develop programs that inform, include, and advance adults with cystinosis, get started at bit.ly/39jVQ3J.

Current ALAB initiatives include:

CystinosisTEENS. A private Instagram account dedicated to supporting teens living with cystinosis.

Cystinosis Rare: A Journey Into the Unknown. A podcast series covering topics impacting our adults with cystinosis and the overall community. Subscribe to the CRN

YouTube channel to receive new video alerts. To date, subject matters have included; coping mechanisms, success stories, loss and grief, transition, bullying, pandemic stress/ anxiety and COVID-19 vaccines.

Cystinosis Sessions. Live, face-to-face (video) conversations with cystinosis stakeholders. Sessions are led by adults with cystinosis but all are invited to participate. Topics have included: COVID-19, medication, independence, education, employment, overcoming limitations & goal setting, mental health, exercise,

and more. Visit the ALAB page url listed below or the cystinosis.org events page for the latest information on these quarterly events.

Cystinosis Memorial Fund. Apply for financial support to cover technology, career and tutoring/educational needs, prevention of muscle wasting and more. Open to teens and adults living with cystinosis. Learn more at cystinosis.org/cmf.

For additional information on the Adult Leadership Advisory Board and current members (listed below), visit: cystinosis.org/support-resources/alab.

ALAB Members



*Jana Healy
Chairperson*



*Steve Schleuder
Vice Chairperson*



Briana Dundon



Brian Ensor



Karen Gledhill



Sara Healy



Christina Morris



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

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

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

How you can help

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

- 1. Go to allstripes.com/cystinosis**

- 2. Create your account**

Sign up, review our research consent and share the names of your health facilities (about 10-15 minutes). No appointments, no uploads.

- 3. Success — you're a hero for cystinosis**

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



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Research Committee Update

By Christy Greeley, Vice President of Research



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

CRN has funded over \$5.5 million total in research grants and fellowship, including a Cystinosis fellowship at the National Institutes of Health, research and education programs in the United States and many countries around the world including Egypt, Mexico, England, Scotland, Italy, Belgium, France, and much more. CRN has also co-funded research projects with Cystinosis Ireland and does so currently. CRN research topics have focused on every aspect of cystinosis with the purpose of understanding the disease and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, fertility, improved medications, psychological and much more. CRN's current grant commitment is approximately \$800,000 covering six different research projects, including one co-funded with Cystinosis Ireland.

NEWLY FUNDED RESEARCH PROJECTS

CRN is pleased to announce funding of the following new research project:

Development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis, Drs. Katharina Hohenfellner and Julia Quitmann

Total project investment: \$155,075.09, two years

Patient-Reported Outcome measures (PROM) are questionnaire-based tools that can help healthcare professionals understand the health status or disease burden from the patient's perspective. These tools can be used to evaluate new therapies or to improve the healthcare provided. Disease-specific instruments that measure health-related quality of life (HrQoL) are particularly informative, as they capture the needs and challenges of specific patient groups particularly well. As a multidimensional construct, HrQoL includes physical, emotional, mental, social, and behavioral components of well-being from the patient's perspective. HrQoL can be measured using four different types of instruments: generic, chronic-generic, condition specific

and treatment-specific instruments. Generic questionnaires represent the full range of health conditions, address groups independent of their respective health state and are effective for comparisons between two cohorts (e.g. patients with cystinosis and healthy controls). Chronic-generic instruments are focusing on a chronic condition independent of its specific characteristics, while specific questionnaires are tailored to problems associated with a specific condition (e.g. cystinosis) or treatment (e.g. patients receiving a kidney transplantation). Despite the significant impairments experienced by patients with cystinosis, very few studies investigate HrQoL in this patient group and disease-specific HrQoL measures are lacking. Thus, the primary aim of this planned study is to develop a PROM for children and adolescents with cystinosis. This instrument will capture the HrQoL from both the child/adolescent and parent perspectives. It will be applicable to clinical trials ranging from randomized clinical trials (RCTs) to surveillance designs, focusing on the impact of cystinosis and its treatment. The preparations have already started. We are currently developing the questionnaire "QUALIFY" (Health-related quality of life of children and adolescents with cystinosis) through intensive literature research and interviews with young German patients and their parents. This preliminary version of QUALIFY needs to be cross-culturally validated in a larger sample (= investigated whether the instrument actually measures what it is supposed to measure) and adapted to the English, Spanish

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

CURRENT CRN GRANT COMMITMENTS

- Cognitive Control Systems in Cystinosis Sophie Molholm, PhD Co-Principal Investigator, John Foxe, PhD Co-Principal Investigator Grant Amount: \$315,193, two-year study

- Chitotriosidase as a Therapeutic Monitor for Cysteamine Therapy in Cystinosis: A Retrospective Validation Study, Mohamed A. Elmonem, Koenraad R.P. Veys, Lambertus P. van den Heuvel, William A. Gahl, Elena Levchenko Grant amount: \$44,000, one year study

- IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis, Katharina Hohenfellner, MD Grant Amount: \$77,000, one year study

- Perturbations in the V-ATPase Pathway Drive Pathology in the Male Reproductive System in Cystinosis, Grant Awarded February 2021 by the Cystinosis Research Network and Cystinosis Ireland, 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: €300,000 from Cystinosis Ireland and CRN (€150,000 each) over the next three years

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

of Pediatric Nephrology, Leuven, EU, and Swastika Sur, MSc., PhD Postdoctoral Scholar, Sarwal Lab, University of California San Francisco, Department of Surgery: Total Grant: €10,000

RESEARCH UPDATES

Cognitive Control Systems in Cystinosis, Ana Francisco, Sophie Molholm, PhD, John Foxe, PhD

The past two years have been challenging for most of us in many ways. The world paused. And so did the Cognitive Neurophysiology Lab. We didn't know if we would be able to make up for the lost time, but we were not willing to risk our participants' and their families' health, so we waited.

SPOILER ALERT: We're back!

While we were waiting, we did our best to stay in touch with the community and thought of ways to understand the impact that the pandemic was having on the cystinosis and other communities with rare genetic conditions. For example, we created an online survey that asked questions about feelings, experiences, fears, and thoughts about things like vaccination. A quick look at your answers tells us that changes in access to medical care (less communication and appointments) further strained the community. During 2021, increased fear and anxiety- and depression-type symptoms were more common and burdensome. And, due to the needed prolonged social isolation, most families had concerns about future social interactions and how successful their children would be when resuming regular life. Though most were willing to get vaccinated, there were serious concerns about the lack of knowledge and evidence on the potential effects of the COVID-19 vaccine in those living with a rare condition.

If you or a family member are struggling, please consider looking for help. If you don't know how or where to start, contact us (Ana Francisco, ana.alvesfrancisco@einsteinmed.edu or 718-862-1824) and we'll give you our best advice on how to look for adequate services.

While we were waiting, we also took the time to prepare our new project. The goal of this new project is to characterize cognitive control in cystinosis, focusing on different components and skills. Briefly, cognitive control is the ability to engage in goal-oriented behaviors, allowing the brain to solve difficult, novel, or complex tasks, such as correcting errors. We're particularly excited about this project, because it was developed after conversations with families and people with cystinosis, who feel like so many of their difficulties are cognitive control difficulties. In this new project we use EEG (remember the funny swimming caps and the many electrodes which allow us to look at your brain activity?), cognitive testing, and questionnaires. And we'll relate all these measures to get at the bigger picture and hopefully use science to explain those difficulties felt by people with cystinosis and contribute to the development of target interventions, leveraging strengths and working on weaknesses. In this project, we are also adding a group of parents. This will allow us to better test what the impact of the mutation versus the impact of the disease is on the cognitive profile associated with cystinosis (also, the parents' inclusion might or might not have been a request from our participants with cystinosis, who felt the parents ought to do some work too J).

So, last year we were busy creating



Jack and Alex Greeley.

and testing the new computer tasks (really fun this time around!) to make sure they were working well and were adequate for many different ages. We have rocked those funny EEG caps too many times in your absence... And now, with a fully vaccinated staff and taking all precautions, we have started welcoming participants and their families! Recruitment has begun and we had our first visits in December and January. But we need many more of you! So, if you or your child have cystinosis and you'd like to come to New York to participate in a research study, contact Ana Francisco at ana.alvesfrancisco@einsteinmed.edu or 718-862-1824. This is a 3-day study involving in-person visits to our lab. We cover accommodation and travel and will give you the best tips about New York.

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis Project report summary, Dr. Katharina Hohenfellner

In patients with cystinosis the condition of the musculature is important for the course of the

disease. Regular exercise builds muscle and can positively influence life expectancy and quality of life. IMPACT aims to show whether regular exercise with vibration plates increases muscle strength in cystinosis patients, to reduce serious medical interventions in the long term and to improve quality of life. To do so, the study uses a training concept, based on the established approach of pediatric rehabilitation "Auf die Beine", developed for children and adolescents with limited mobility, where participants exercise while standing on a Galileo vibration platform. For comparison a second group exercises only their arms using a vibration dumbbell. Both exercise according to a fixed training schedule with 10 short training sessions per week. Each training session lasts between 5 and 8 minutes. The training program is tailored to the disease, and the dumbbell exercises are simplified for the younger children. Training at home and the short duration aims to facilitate integration into everyday life. To assess the concept, patients

are asked to document their training progress as well as any difficulties or side effects. During the exercising period patients are contacted via video to check on training progress and to address potential problems. In addition, the training is accompanied by a baseline and three follow-up clinical assessment, one after the three-month training phase, one after a three-month follow-up phase, and one after one year. Participants are provided with step counters to measure daily activity and a quality-of-life questionnaire before each assessment. During the assessments muscle strength and cardio-respiratory function are measured. Additionally, at baseline and after one year, a physiotherapeutic and orthopedic examination and routine clinical and laboratory evaluations are performed. Twenty-four adult patients from Germany and Italy, participated in the Study between September 2020 and September 2021. In addition, fifteen juvenile patients (between 5 and 12 years) from Germany, Austria, and Slovakia started their study in September 2021 and will finish in September 2022. Based on sex, age, and kidney transplant status, patients were randomized into the group using the vibration platforms are the one using the vibration dumbbell. The first analysis of the training diaries shows, that 20 out of the 23 adult participants, who completed the training period, completed at least 80% of the required 10 training sessions per week, 15 of them even more than 90%. The adult participants reported no negative effects during the training sessions (injuries or pain due to the training). Out of the 14 children, who completed the training period, 11 met at least 80% of the required training sessions, 9 met more than 90%. Even though they performed simplified

exercises and used dumbbells, designed for children, the training with the dumbbell was difficult for 4/15 children. Parents also had to help more with the exercises with the dumbbells than with the platforms. Nonetheless, parents of both groups gave positive feedback and repeatedly reported visible improvements in their children. The results of the comprehensive statistical analysis for the adult group are expected in June 2022, results for the juvenile subgroup at the end of the year.

CYSTINOSIS COMMUNITY ADVISORY BOARD

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

This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research. We look forward to continued partnership with researchers and industry worldwide to improve the quality and speed with which cystinosis treatments are developed with the patient's voice in mind. The CAB will meet in person for the first time in nearly three years in conjunction with the Cystinosis Network Europe meeting in Brussels in July.

NATIONAL INSTITUTES OF HEALTH

As a reminder, patients may contact the National Institutes of Health to be enrolled in the cystinosis protocol and for consultative care. For more information, please contact: Joy Bryant, (301) 443-8690, bryantjo@mail.cc.nih.gov

EDUCATIONAL RESOURCES

All of CRN's educational materials including brochures, guides and other publications have been updated and are available on the CRN Website. Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from around the world. Also be sure to check out the many cystinosis related articles and publications available in our Publications and Guides library at Publications & Guides (cystinosis.org)

RESEARCH PARTICIPATION OPPORTUNITIES

Clinical researchers from the Stanford Neuromuscular department are looking for individuals who have been diagnosed with Cystinosis Myopathy, to participate in a high-intensity interval training (HIIT) exercise study. Further information about this study is listed on the clinicaltrials.gov website here.

There is no cost to participate in the study, and as part of your participation in this study you will receive an exercise bike delivered to your home, free of charge, that you get to keep after study cessation. We are looking for participants who currently reside in the USA. Unfortunately, we cannot accept participants living outside the USA due to limitations in shipping.

If this is something you may be interested in, please email cfrater@stanford.edu for further details.

HAVE YOU OR YOUR CHILD BEEN DIAGNOSED WITH CYSTINOSIS?

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

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AVROBIO Reports Positive Data from Phase 1/2 Clinical Trial of Investigational Gene Therapy for Cystinosis, including New Interim Data on Neurocognitive Measures

New early data show key visual motor integration, visual perception and motor coordination measures impacted by cystinosis stabilized or improved post gene therapy

Systemic reach of AVR-RD-04 observed across multiple other measures

All five dosed patients remain off oral cysteamine post gene therapy

Continued favorable safety profile with no adverse events related to drug product to date

Clinical proof-of-concept in adult patients lays groundwork for AVROBIO-sponsored clinical trial planned to begin in 2023

CAMBRIDGE, Mass.—(BUSINESS WIRE)—May 17, 2022—AVROBIO, Inc. (Nasdaq: AVRO), a leading clinical-stage gene therapy company with a shared purpose to free people from a lifetime of genetic disease, today reported new interim data, including on new visual motor integration, motor coordination and visual perception measures, from a collaborator-sponsored, ongoing Phase 1/2 gene therapy clinical trial of AVR-RD-04, an investigational gene therapy for cystinosis, at the 25th Annual Meeting of American Society for Gene and Cell Therapy (ASGCT) in Washington D.C., May 16-19, 2022.

The collaborator-sponsored Phase 1/2 clinical trial is evaluating the safety and efficacy of AVR-RD-04 in adult patients diagnosed with the infantile form of cystinosis who previously had been treated with the current standard of care (SOC) cysteamine. AVR-RD-04 genetically modifies patients' own hematopoietic stem cells (HSC) to express a functional version of cystinosin, the protein that is deficient in people living with cystinosis. Preliminary data suggest that post gene therapy, functional cystinosin has been produced throughout

the body as evidenced by clinical measures in multiple tissues, including the eyes, skin, gastrointestinal mucosa and neurocognitive system. No adverse events (AEs) related to the drug product have been reported to date.

"We're thrilled with our progress in this first and only gene therapy trial for cystinosis, a devastating genetic disease with unmet medical needs that impact the daily lives of patients and their families," said Stephanie Cherqui, Ph.D., lead investigator of the clinical trial and associate professor of Pediatrics at the University of California San Diego (UCSD). "Now with data from up to five patients, we have observed a strong safety and tolerability profile, as well as a reduction in the harmful accumulation of cystine crystals in cells across multiple tissues.

"All five patients dosed to date remain off oral cysteamine. We believe the results to date for this investigational gene therapy show its potential to stabilize or reduce impact of cystinosis on different tissues with a single dose," she added.

The collaborator-sponsored Phase 1/2 clinical trial of AVR-

RD-04 is funded in part by grants to UCSD from the California Institute for Regenerative Medicine (CIRM), Cystinosis Research Foundation (CRF) and National Institutes of Health (NIH).

"We believe the interim data from this ongoing clinical trial demonstrate the potential of gene therapy using patient's own hematopoietic stem cells to impact the body head-to-toe by restoring functional cystinosin and reducing the accumulation of cystine crystals systemically," said AVROBIO Chief Medical Officer, Essra Ridha, M.D., MRCP, FFPM. "Cystinosis is a devastating disease that currently carries a 5-year treatment cost in excess of \$4 million per patient in the U.S. and impacts approximately 1,600 patients in the U.S., Europe and Japan alone. With proof-of-concept demonstrated, we continue to lay the groundwork for an AVROBIO-sponsored clinical trial planned to begin in 2023 and look forward to our interactions with regulators on our clinical and Chemistry Manufacturing and Controls (CMC) strategy later this year."

Key motor coordination and visual perception measures stabilize or show

positive trends post gene therapy

Visual motor integration (VMI) measured with the Beery – Buktenica Developmental Test of VMI, a standardized test evaluating the ability of the brain to interpret and translate visual information into an exact motor response, has been shown to be a consistent indicator of visual spatial and visual motor dysfunction in patients with cystinosis. These measures do not generally improve over time in this population.

Early data indicate that post gene therapy the two patients with data to date show stabilization of scores on the Beery – Buktenica Developmental Test of VMI and importantly, improvement in the subtests of motor coordination and visual perception, suggesting a potential impact on the neuropathology of the disease. In patient #1, an approximate 20-point improvement was evident in both visual perception and motor coordination, and in patient #3 a 5-point increase in visual perception was detected, with motor coordination rising by 45 points in the first 6 months post treatment and a more modest rise thereafter.

In addition, following discontinuation of cysteamine, average hand grip strength remained stable up to 27 months after dosing.

Systemic reach of AVR-RD-04 also seen across measurements of blood, eye, skin and gastrointestinal mucosa

Early data indicate that post gene therapy, patients have been able to produce and distribute functional cystinosis protein throughout the body, which prevents the pathological accumulation of cystine crystals. In blood, the leukocyte cystine levels decreased measurably, with the three patients out more than 12 months post gene therapy stabilizing near 1.0

nmol/mg protein.

Photophobia, or extreme visual sensitivity to light, is a hallmark of cystinosis. In a patient-reported outcome scale of photophobia severity, the first three patients for which data are available, reported improved or stable photophobia scores. Patient #1, who entered the trial with a higher level of cystine crystal accumulation in the eye, reported a two-point photophobia score improvement 24 months post gene therapy. Patients #2 and #3, who both entered the trial with relatively lower cystine crystal accumulation in the eye, reported stable photophobia scores, both at 12 months post gene therapy. Patients #1, #3, #4 and #5 remain off cysteamine eye drops.

A decline in cystine crystals was observed in skin and gastrointestinal mucosa biopsies from the first three patients. Patients with cystinosis accumulate cystine crystals in cells, which leads to tissue and organ damage and results in debilitating co-morbidities. In the skin, reductions in average intracytoplasmic crystals per cell ranged from 8% in patient #1, 64% in patient #2 and 81% in patient #3 below the patients' own standard-of-care baseline measures at 12-27 months post gene therapy. In the intestinal mucosa, a measurable reduction below patients' own standard-of-care baseline measures was observed post gene therapy, including for patient #1 a 73% reduction after 27 months, for patient #2 a 28% reduction after 12 months and for patient #3 an 83% reduction after 18 months. These data suggest the systemic distribution of functional cystinosis protein is impacting a variety of measures throughout the body. Biopsies have not yet been conducted for patient #4 and #5, who have been more recently infused.

Darker pigmentation observed may be a sign of multi-functional cystinosis activity post gene therapy

Patients with cystinosis frequently exhibit blond or lighter-colored hair and fair complexion because of reduced levels of melanin in their skin. In vitro studies have demonstrated that cystinosis is located in melanosomes of melanocytes and when functional cystinosis is absent or reduced, melanin pigment synthesis is inhibited.

New early quantitative data suggest that gene therapy-derived cystinosis may restore melanin production. Twelve months after infusion, two patients exhibited progressively darkening hair color, as measured by a 25% and 37% reduction in red, green, blue (RGB) mean intensity for patient #1 and patient #3, respectively, further indicating cystinosis protein throughout the body. In this case, a microscope was used to obtain high resolution images of hair strands. The images were taken using transmitted light at 20x magnification and analyzed for RGB intensity with numerical values assigned to quantify the level of pigmentation. These data are not yet available for patients #2, #4 and #5.

Sustained engraftment demonstrated with stable VCN for patients beyond 12 months

Importantly, sustained engraftment has been observed in the first three patients, as evidenced by stable vector copy number (VCN) levels. At 17- to 27-months post gene therapy, their VCN is between 1.0 and 2.0 per diploid genome. The recently dosed fourth and fifth patient have a VCN of 0.7 and 1.3 per diploid genome at three-months and one-month post gene therapy, respectively.

Safety and tolerability profile remains

strong

Safety data from the five patients dosed to date indicate no AEs related to drug product. All AEs were related to myeloablative conditioning, stem cell mobilization, underlying disease or pre-existing conditions. The majority of AEs were mild or moderate and resolved without clinical sequelae.

Further details on the collaborator-sponsored Phase 1/2 trial (NCT03897361) are available on clinicaltrials.gov. For more information on the cystinosis trial data presented at ASGCT, please see data slides here.

AVROBIO-sponsored trial on track for 2023

Based on the data reported today, combined with feedback provided by the U.S. Food and Drug Administration (FDA) after a fall 2021 Type C meeting, and pending the outcome of further planned regulatory authority interactions this year, the company expects to initiate an AVROBIO-sponsored trial in 2023 in the U.S., followed by sites in the UK and Europe. AVROBIO's current plan involves a two-part strategy, beginning in a pre-renal transplant population followed by a post-renal transplant population.

About cystinosis

Cystinosis is a rare, progressive disease marked by the accumulation of cystine in cellular organelles known as lysosomes. This buildup causes progressive organ damage and debilitating corneal damage, swallowing dysfunction, chronic kidney disease leading to end-stage renal disease and muscle wasting leading to a shortened lifespan. Currently, more than 90% of treated cystinosis patients require a renal transplant in the second or third decade of life. The current standard of care for cystinosis

is cysteamine, a treatment regimen that can require dozens of pills per day, does not prevent overall disease progression and carries side effects, such as breath and body odor and gastrointestinal complications, which may be difficult to tolerate.

About AVROBIO

Our vision is to bring personalized gene therapy to the world. We aim to prevent, halt or reverse disease throughout the body with a single dose of gene therapy designed to drive durable expression of therapeutic protein, even in hard-to-reach tissues and organs including brain, muscle and bone. AVROBIO's pipeline is powered by our industry-leading plato® gene therapy platform, our foundation designed to deliver gene therapy worldwide. It includes clinical programs in cystinosis and Gaucher disease type 1, as well as preclinical programs in Gaucher disease type 3, Hunter syndrome and Pompe disease. We are headquartered in Cambridge, Mass. For additional information, visit avrobio.com, and follow us on Twitter and LinkedIn.

Forward-Looking Statements

This press release contains forward-looking statements, including statements made pursuant to the safe harbor provisions of the Private Securities Litigation Reform Act of 1995. These statements may be identified by words and phrases such as "aims," "anticipates," "believes," "could," "designed to," "estimates," "expects," "forecasts," "goal," "intends," "may," "plans," "possible," "potential," "seeks," "will," and variations of these words and phrases or similar expressions that are intended to identify forward-looking statements. These forward-looking statements include, without limitation, statements regarding our

business strategy for and the potential therapeutic benefits of our product candidates, including AVR-RD-04 for the treatment of cystinosis, the design, commencement, enrollment and timing of ongoing or planned clinical trials, clinical trial results, product approvals and regulatory pathways, anticipated benefits of our gene therapy platform including potential impact on our commercialization activities, timing and likelihood of success, the expected benefits and results of our implementation of the plato platform in our clinical trials and gene therapy programs, and the expected safety profile of our investigational gene therapies. Any such statements in this press release that are not statements of historical fact may be deemed to be forward-looking statements. Results in preclinical or early-stage clinical trials may not be indicative of results from later stage or larger scale clinical trials and do not ensure regulatory approval. You should not place undue reliance on these statements, or the scientific data presented.

Any forward-looking statements in this press release are based on AVROBIO's current expectations, estimates and projections about our industry as well as management's current beliefs and expectations of future events only as of today and are subject to a number of risks and uncertainties that could cause actual results to differ materially and adversely from those set forth in or implied by such forward-looking statements. These risks and uncertainties include, but are not limited to, the risk that any one or more of AVROBIO's product candidates, including AVR-RD-04 for the treatment of cystinosis, will not be successfully developed or commercialized, the risk of cessation

or delay of any ongoing or planned clinical trials of AVROBIO or our collaborators, the risk that AVROBIO may not successfully recruit or enroll a sufficient number of patients for our clinical trials, the risk that AVROBIO may not realize the intended benefits of our gene therapy platform, including the features of our plato® platform, the risk that our product candidates or procedures in connection with the administration thereof will not have the safety or efficacy profile that we anticipate, the risk that prior results, such as signals of safety, activity or durability of effect, observed from preclinical or clinical trials, will not be replicated or will not continue in ongoing or future studies or trials involving AVROBIO's product candidates, the risk that we will be unable to obtain and maintain regulatory approval for our product candidates, the risk that the size and growth potential of the market for our product candidates will not materialize as expected, risks associated with our dependence on third-party suppliers

and manufacturers, risks regarding the accuracy of our estimates of expenses and future revenue, risks relating to our capital requirements and needs for additional financing, risks relating to clinical trial and business interruptions resulting from the COVID-19 outbreak or similar public health crises, including that such interruptions may materially delay our enrollment and development timelines and/or increase our development costs or that data collection efforts may be impaired or otherwise impacted by such crises, and risks relating to our ability to obtain and maintain intellectual property protection for our product candidates. For a discussion of these and other risks and uncertainties, and other important factors, any of which could cause AVROBIO's actual results to differ materially and adversely from those contained in the forward-looking statements, see the section entitled "Risk Factors" in AVROBIO's most recent Quarterly Report, as well as discussions of potential risks,

uncertainties and other important factors in AVROBIO's subsequent filings with the Securities and Exchange Commission. AVROBIO explicitly disclaims any obligation to update any forward-looking statements except to the extent required by law.

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¹Collaborator-sponsored Phase 1/2 clinical trial of AVR-RD-04 is funded in part by grants to UCSD from the California Institute for Regenerative Medicine (CIRM), Cystinosis Research Foundation (CRF) and National Institutes of Health (NIH).

Dr. Gahl Lifetime Achievement

The EURORDIS Lifetime Achievement recognises the lifelong dedication Dr. Gahl has shown in addressing the needs of people living with a rare and undiagnosed disease. With this award, we would like to acknowledge the key role he has played in the creation of the Undiagnosed Diseases Network (UDN) within the National Institutes of Health, a programme that has played an essential role in discovering many new genetic disorders. His unwavering support of the rare and undiagnosed disease cause and his patient-centred approach is demonstrated by the leadership role he played in the development of the

International Network on Undiagnosed Diseases (UDNI). The programme provides an admirable example of an international institution which brings together the undiagnosed community at the global level and creates a structure and forum to exchange on undiagnosed cases for clinicians working worldwide. Additionally, this award also recognises the discoveries made by Gahl's lab of the genetic bases of gray platelet syndrome, Hartnup disease, arterial calcification due to deficiency of CD73, 3-methylglutaconic aciduria type III, 3 types of HPS, and neutropenia due to VPS45 deficiency.



Originally published on eurordis.org.

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

By Tim Wyman, Treasurer

The Cystinosis Research Network (CRN) 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. The CRN is a tax-exempt organization granted "501(c)(3)" non-profit status by the I.R.S. The CRN

Federal tax id # is 04-3323789.

Total income for the 5 months in 2022 was \$289,856 which consisted mainly of \$268,400 in grants received and \$19,252 from fundraising. Expenses totaled \$309,092 for -\$19,236 net income year to date. The majority of expenses were \$239,462 towards research grants (cystinosis.org/research) and \$35,441 to education.

CRN also paid out \$7,200 in scholarships (cystinosis.org/support-resources/scholarships) in the first five months of 2022. Thanks to grants and fundraising by many in the cystinosis community, CRN's current equity (assets minus liabilities) stands at a healthy \$677,759 which is critical in funding additional research.



Celebrating Mothers

By Jessica Magnus

They changed your life when they came into this world. You were forced to grow up inside and be responsible. They allowed you to understand that your words have power when it comes to education, that your caresses were important, that your hugs helped every day to grow a child who feared the dark, who longed to share time with you.

We could say that raising a child also requires us to educate ourselves. We always look for the best for our children, and therefore, we measure our actions and create environments conducive for them to grow up happy. We learn about nutrition, psychology and medicine. We are castle builders and design doll clothes, we are confidants, tear wipers, we have

magic to erase nightmares and monsters from under the bed, and we become chefs of our kids' favorite desserts.

Kids grow up every day, but we grow with them too. Because they are our weakness, because they are our heart outside the body and the breath of our hopes.

There are no words to express my admiration for those moms I have been able to meet over the years.

To them who have to live the hardest part of motherhood, sleeping in a chair in a hospital next to the beings they love most. To them who have learned by force to get up off the floor and move on for their children... Super Moms.



Nephropathic Cystinosis: Apply for Assistance

Good Days provides financial support for patients who cannot afford the treatment they urgently need. Good Days has streamlined the enrollment process so patients can receive immediate determination of eligibility for financial assistance.

Eligibility Criteria

- Patient must be diagnosed with a covered disease and program must be accepting enrollments
- Patient must have a valid Social Security number to apply for assistance and receive treatment in the United States
- Patient must be seeking assistance for a prescribed medication that is FDA approved to treat the covered diagnosis
- Patient is required to have valid insurance coverage
- Patient income level must meet program guidelines

Enrollment Checklist

1. Patient contact and demographic information
2. Health insurance information
3. Diagnosis and medication
4. Prescribing physician's name and phone number
5. Estimate of patient's household income and household size

Get started at mygooddays.org/apply.

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

CRN VISION

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

CRN MISSION

The Cystinosis Research Network 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.