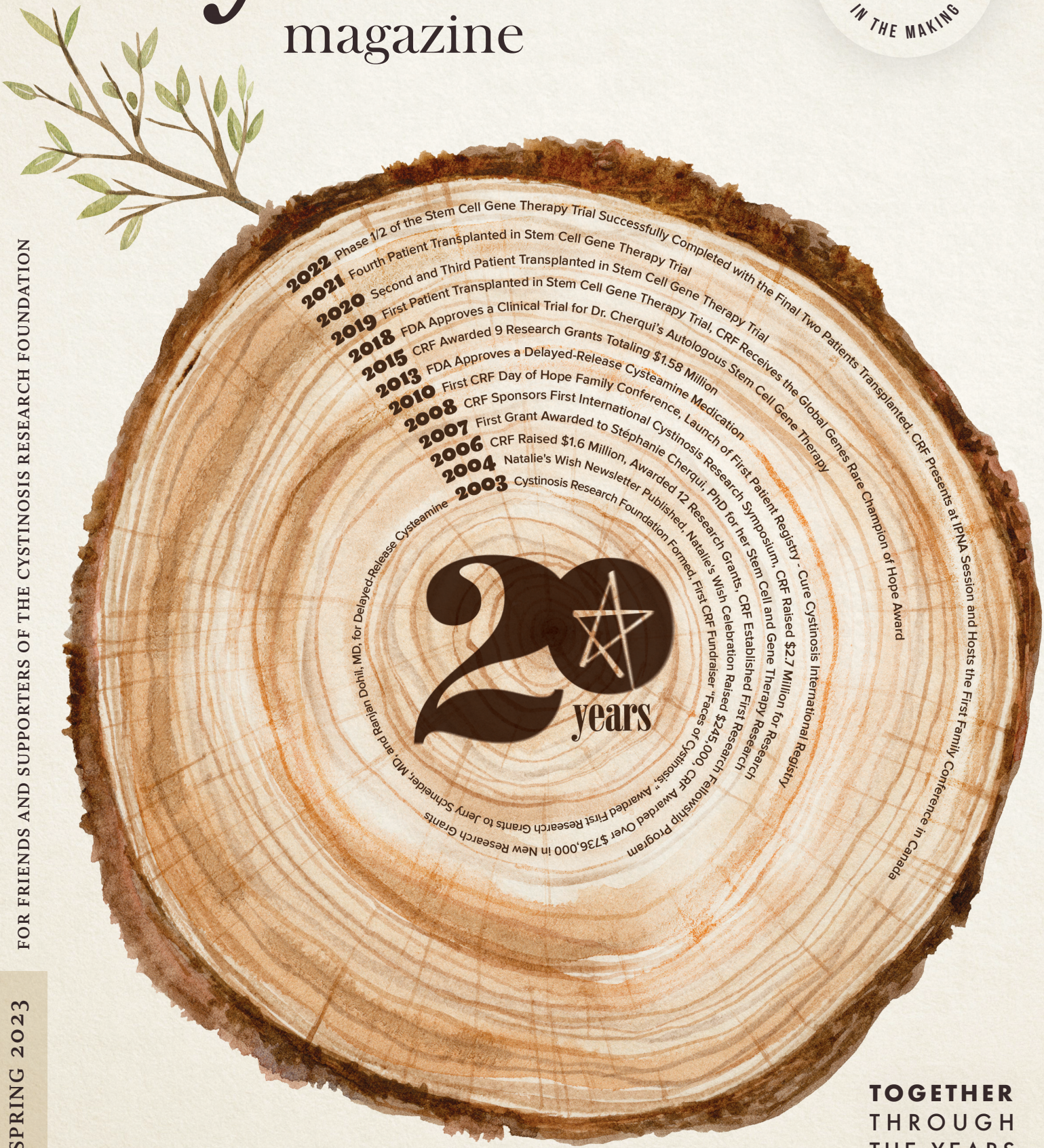
# cystinosis magazine

OUR HISTORY IS A **VICTORY** IN THE MAKING




**TOGETHER THROUGH THE YEARS**

FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

SPRING 2023

**2003**

- Natalie Stack made a wish on the eve of her 12th birthday, "to have my disease go away forever."
- The Cystinosis Research Foundation was established with the sole purpose of raising funds to find better treatments and a cure for cystinosis.

**2008**

- First CRF International Research Symposium

**2013**

- FDA approval in 2013 for a delayed-release form of cysteamine. CRF funded every early clinical study that led to the discovery of the delayed-release form of the medication now known as Procysbi®.
- First patient pilot study for an allogeneic stem cell study at UCLA.

**2018**

- FDA approval on December 19, 2018 for first stem cell and gene therapy clinical trial to test a new treatment for cystinosis.

**2019**

- First patient in stem cell and gene therapy clinical trial transplanted on October 7, 2019.

**2020**

- Second patient in stem cell and gene therapy clinical trial transplanted on June 29, 2020.
- Third patient in stem cell and gene therapy clinical trial transplanted on November 16, 2020.

**2021**

- Fourth patient in stem cell and gene therapy clinical trial transplanted on November 15, 2021.
- CRF partnered with Sanford CoRDS to create the new Cure Cystinosis International Registry *(CCIR),* the only international cystinosis patient registry in the world.

**2022**

- Fifth patient in stem cell and gene therapy clinical trial transplanted on March 29, 2022.
- Sixth patient in stem cell and gene therapy trial was transplanted October 24, 2022.
- CRF Presents at IPNA pre-Congress Cystinosis Session in Calgary, Alberta, Canada and hosts the first Family Conference.

20 years

SPRING 2023

## 20 YEARS DEEP

Strong roots
produce strong trees.
We grow stronger
in community.

The tree rings
reveal our history
as we all grow
in the CRF story.

Our dedication.
Our connection.
Our cystinosis journey

**TOGETHER**
through the years.

**CONTACT US:**
Please send suggestions and comments regarding *Cystinosis Magazine* to *nstack@cystinosisresearch.org.*

To receive our e-newsletter, *Star Facts*, send your email address to *zsolsby@cystinosisresearch.org.*

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CYSTINOSISRESEARCH.ORG

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised over $67 million for cystinosis research in an effort to find a cure.

cystinosis RESEARCH FOUNDATION

## WELCOME





## RESEARCH NEWS



## ANNOUNCEMENTS






## FAMILY STORIES




*CYSTINOSIS MAGAZINE* IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

**"With gratitude, optimism is sustainable."**

MICHAEL J. FOX

# Dear Family and Friends,

SPRING 2023

We are so grateful for your steadfast support throughout the years. You have embraced our community, supported our research efforts, and have stood by our side as we seek the cure for cystinosis. Your support has fueled our optimism and allowed us to remain hopeful when times have been challenging.

In some ways, it seems like only yesterday that Natalie, just days away from her twelfth birthday, made her wish, "to have my disease go away forever". When Natalie was diagnosed in 1991, we were told she would not live long enough to graduate from high school. The diagnosis left us feeling alone and isolated. It was difficult to feel hopeful because of the lack of resources, community and research. When Natalie made her wish, everything changed and hope became our mantra.

Twenty years ago, after Natalie wrote her wish on a napkin, we established the Cystinosis Research Foundation. We have since grown into a global research powerhouse that has changed the way we think about cystinosis. We have built a global scientific community one study at a time, and today we have researchers in 12 countries working on better treatments and a cure for cystinosis.

We have funded research that has led to two FDA approvals and we are on the brink of other clinical trials that will improve the lives of those with cystinosis. In twenty years, we have built a strong foundation for innovative research that is not only thriving, but producing extraordinary results.

Natalie's wish transformed our lives in the most profound and significant ways. The devastating diagnosis of cystinosis unexpectedly and gratefully brought us many blessings. Every milestone and accomplishment we have made is because of you! We are no longer isolated or hopeless – you have become our family, our community. We are so grateful for your partnership; we have so much to be thankful for because you have been an integral part of our community's journey.

20 years

## CYSTINOSIS; A CHRONIC AND LIFE-THREATENING DISEASE

Cystinosis is a progressive and life-threatening disease. Cystinosis destroys every organ in the body including the kidneys, eyes, liver, muscle and brain. With each decade of life, there are more complications from the disease including kidney transplants, bone issues, muscle wasting and myopathy, neurological issues and ocular deterioration. Most patients with cystinosis take between 9-12 different medications every day just to stay alive; there are no days off from cystinosis. Although there is a medication that prolongs the lives of our children and adults with cystinosis, it is not a cure. We have more work to do and must continue to seek better treatments to improve the lives of those living with cystinosis.

## THANK YOU FOR DONATING A REMARKABLE $1,140,100 DURING NATALIE'S WISH IN APRIL!

Together, we have created a thriving, global research community, and with your help, we will continue to fund research aimed at developing new treatments and finding a cure for cystinosis. Since 2003, CRF has awarded 223 research grants, eight extension grants and nine equipment grants in 12 countries. Our global research teams have made discoveries that have led to significant advances in the treatment of cystinosis and new discoveries about cystinosis and its complications.

CRF awards research grants twice a year, ensuring there is never a gap in the research cycle and that donations are being put to work throughout the year. We often fund researchers with new ideas, giving them the opportunity to successfully test their theories and the ability to seek grants from other funding sources, leveraging CRF's initial grant awards. Our strategic approach to research and our emphasis on collaboration has accelerated research and has expanded the field of cystinosis. CRF funded researchers have published 103 articles in prestigious research and medical journals. Every dollar you donate goes directly to research to fund the best and the brightest researchers in the world.

## RESEARCH GIVES US HOPE

### NINE RESEARCH GRANTS AWARDED IN 2022 TOTALING $1,608,127

CRF has built a synergistic, collaborative international research community that is working around the clock to find better treatments and a cure for cystinosis.

We are pleased to announce that in 2022, we awarded nine new grants, totaling $1,608,127. The grants were awarded to researchers in France, Germany, Italy, New Zealand, Switzerland and the United States. The research awards include areas of study about bone disease, ocular cystinosis, novel medications and kidney organoids. We have a comprehensive list of all researchers we have funded and the total number of grants issued in specific areas of cystinosis research beginning on page 66. You will find descriptions of the two most recent grants awarded in December 2022 on page 71.

We are thankful to the CRF research community for their unwavering commitment to our community and for giving us hope with every discovery they make that moves us one step closer to the cure.

The success of the CRF research program is a result of the leadership and commitment of the CRF Scientific Review Board (SRB), chaired by Corinne Antignac, MD, PhD. The SRB reviews and critiques the merits of every research application received during our spring and fall call for new applications. As a result of our biannual grant award program, CRF is the leading fund provider of cystinosis research grants in the world.

We are eternally grateful to the nine members of the CRF SRB, listed on page 75, for their dedication to our community and our research program. They have been instrumental in shaping the direction of the research program. We simply could not do any of this without their expertise, guidance and leadership.

## DID YOU KNOW? CYSTINOSIS RESEARCH HELPS OTHER DISEASES

We set out to find better treatments and a cure for cystinosis. Along the way we have learned that not only have we made significant advances in the field of cystinosis, but the research we fund has had a positive impact on other more prevalent diseases and disorders. Discoveries made by CRF-funded researchers are being applied to other disease groups including Friedreich's Ataxia, Danon disease, corneal diseases, kidney diseases and genetic and systemic diseases similar to cystinosis. A cure for cystinosis will help find cures for other diseases, potentially helping millions of people. We are changing lives together.

## GOOD NEWS ON THE STEM CELL AND GENE THERAPY TRIAL

As reported late last year, the sixth and final patient in this phase 1/2 of the FDA approved stem cell and gene therapy clinical trial was transplanted. When I reflect on the time it took to get FDA approval for the trial, I am astounded by the relatively short timeline. In 2007, CRF awarded the first of many grants to Stéphanie Cherqui, PhD, for her stem cell and gene therapy work. Eleven years later, in 2018, the FDA approved a clinical trial to test her treatment! That is a remarkable feat by anyone's standard.

CRF has awarded over $6.1 million in grants to Dr. Cherqui for her research. Our early funding allowed Dr. Cherqui to test her theories and collect enough preliminary data to present to other, larger funding agencies. Dr. Cherqui has secured over $21.5 million in additional grants from other funding agencies including CIRM and the NIH for the stem cell and gene therapy treatment. It was CRF's seed money that launched this life-changing therapy that we hope will be a cure.

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The trial would not be a success without the adult patients who bravely volunteered to be the first patients in the stem cell and gene therapy trial. A heartfelt thank you to Jordan Janz, Jacob Seachord, Tyler Joynt, Natalie Stack and Kurt Gillenberg, who have openly and generously shared their stories with all of us. We have learned so much from them about optimism and hope, coping with uncertainty and gratefulness. They are pioneers who have helped advance cystinosis research and have given us hope that one day soon this treatment will be available to all children and adults with cystinosis. We are so very pleased to report that all the patients are doing very well and remain off oral cysteamine treatment.

We anticipate that the next phase of the clinical trial will commence later this year, which will allow more patients with cystinosis to have the treatment. This is an exciting and hopeful time for the cystinosis community.

## LAUGHTER, JOY AND CELEBRATION – CYSTINOSIS FAMILIES UNITE

Our family conference roared back to life in March as we gathered in Newport Beach, California. Over 260 people attended the CRF Day of Hope family conference. It was truly a CRF family reunion where smiles were broad, hugs were abundant and love and kindness were in the air!

The conference featured several CRF researchers who updated the community about their research projects. Presentations included areas of research and treatment that are most important to cystinosis families, including kidney disease, neurological issues, bone and muscle disease, ocular cystinosis, potential new treatments and current clinical trials, including an update on the stem cell and gene therapy trial.

The conference agenda featured two families and one adult with cystinosis who shared their personal stories about living with cystinosis. Their presentations were the highlight of the conference. The stories reminded us that although we all have a unique cystinosis story, we also share a lot of the same fears, joys and dreams.

We know you will enjoy reading about the conference on page 8 and we guarantee you will smile when you see the pictures of our community together.

## LOOKING AHEAD

This issue of *Cystinosis Magazine* promises to warm your heart with new and updated family and patient stories. The CRF community is extraordinary in every way. Their honesty and openness about sharing their lives with cystinosis is a gift to us. Each story written unites us and reminds us how connected we are and how fortunate we are to have each other.

We have an important message for all of you on page 76. We are going “green” which means that we will not be publishing a hard copy of the magazine starting next year. We want to stay in touch, so please send us your email address so that you receive all the news, research updates and family stories in your inbox.

## WITH HOPE AND GRATITUDE

You have been by our side for 20 years, encouraging us and supporting us in so many ways. We have accomplished milestones once thought to be unimaginable. We have funded studies that have led to two FDA approvals, numerous clinical trials and significant discoveries about cystinosis, all of which have put us one step closer to the cure. We have faced the challenges of cystinosis together, we have overcome obstacles and we have found a path to better treatments and a cure.

You have embraced our community in every way. You are part of the fabric of CRF and we are forever grateful to you for supporting Natalie, our family and our community. Our optimism is sustainable because of your love for our cystinosis community.

With heartfelt thanks and gratitude,

Nancy & Jeff

# A Note from Natalie Stack

I moved to Chicago with my fiancé, Danny, four months ago. It has been quite an adventure! The winter has not been as frigid as most other winters, so I have been able to adapt a little better than expected. We are enjoying our time in a new city and like to go out to try new restaurants every weekend.

Three months ago, I started a new job at Children's Home and Aid which has kept me busy. I am an Adoption Specialist and work with the Illinois Department of Children and Families as a social worker for youth in foster care and with clients who are interested in adopting youth in foster care. The work I do is very rewarding but can also be emotionally exhausting. I am learning something new every day and am excited to learn more as I continue this career path.

It was one year ago in March that I had the stem cell and gene therapy transplant. I am now one year post-stem cell transplant and I continue to feel well. I just completed all the one-year post-transplant exams at UC San Diego. The exams included a multitude of tests that included strenuous and sometimes painful eye exams, mole mapping, skin confocal test, a rectal biopsy, skin biopsy, grip strength test, neurological functioning tests, a 24-hour urine collection, and, of course, multiple blood draws.

I also received five different immunizations which were important since they will help support a healthy immune system. When I had chemotherapy as part of the transplant, it wiped out all the immunizations I received as a baby and child so I have had to be extra cautious about protecting my health. Now, I can breathe a bit easier!

As part of the trial, I will be returning to UCSD later in the year for my 30-month post-transplant testing and will be going through the same extensive testing. Though demanding on my body and mentally exhausting, the tests and the results are an important way to measure the success of the transplant and engraftment. I hope that my results continue to get better and that I get healthier every day. I am so very thankful that I had the opportunity to participate in the study and now have a chance to live a longer life.

I recently flew to California to attend the Day of Hope family conference, which was the first conference I have attended since 2019. It was also the 20-year anniversary of when I made my wish "to have my disease go away forever". It was such a joy to see so many families and children. So many of the younger children that I remembered from the past were now so grown up! It was wonderful to reconnect with the community and hear so many positive updates from the researchers and doctors.

Our community, though small, is filled with love, passion, resilience and determination. It is amazing to witness what our small community has accomplished in only 20 years. We were able to find a delayed-release medication which has improved lives and, more recently, the stem cell treatment which we hope is a cure for cystinosis. I am so grateful to be a part of this community.

I want to thank the CRF community for their determination and dedication to raising money so that we could support research to find better treatments and a cure for cystinosis. Your support made the stem cell trial a reality.

The fight is not over, but I know that our supporters and community will continue to make my wish a reality. We have hope as a community because you have supported research that has changed lives.

Thank you for never giving up and for making my wish — to have my disease go away forever — more certain than ever.

Love,
Natalie

# What is cystinosis?

Cystinosis is a rare, inherited, metabolic disease that is characterized by the abnormal accumulation of the amino acid cystine in every cell in the body. Buildup of cystine in the cells eventually destroys all major organs of the body, including the kidneys, liver, eyes, muscles, bone marrow, thyroid and brain. Medication is available to control some of the symptoms of this terrible disease, but cystinosis remains incurable. Cystinosis affects approximately 600 people, mostly children, in North America, and about 2,500 people worldwide.

**It is one of the 7,000 rare, or "orphan" diseases in the United States that collectively impacts approximately 30 million Americans.**

Federal funding for research on cystinosis and other rare diseases is virtually non-existent and most pharmaceutical companies remain uninterested because financial rewards are too small. Yet, while there are only a small number of patients who suffer from any given "orphan" disease, knowledge gained by studying one disease often leads to advancements in other rare diseases and more prevalent and well-known disorders.

# Our story

In 2003, Natalie Stack made a wish on the eve of her 12th birthday, "to have my disease go away forever." That same year, the Cystinosis Research Foundation (CRF) was established with the sole purpose of raising funds to find better treatments and a cure for cystinosis.

**Today, CRF is the largest fund provider of grants for cystinosis research in the world, issuing 223 grants in 12 countries.**

CRF has raised more than $67 million, with 100% of your donations going to support cystinosis research. CRF's efforts have changed the course of cystinosis and given new energy to its investigators and scientists. CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.


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20 years

cystinosis RESEARCH FOUNDATION
TOGETHER WE SHINE BRIGHT

**We celebrate our CRF community and are grateful every day for your support.**

CRF's highly strategic approach to funding has resulted in two FDA approvals and several human clinical trials. The research dollars we have invested have been leveraged by over $28 million in grants from other funding agencies. Not only does CRF research help our community, but our discoveries are applied to more prevalent diseases and disorders. CRF-funded research has the potential to help millions of others.

We want to thank our families, friends and donors who have remained steadfast in their commitment to finding better treatments and a cure. Thank you to the cystinosis researchers and scientists who are working around the clock on behalf of our children and adults with cystinosis.

*You have changed the course of cystinosis...Thank You!*

*Since 2003, CRF has:*

FUNDED **223** **Multi-Year Grants** in 12 Countries

PUBLISHED **103** **Articles** in Prestigious Journals by CRF Researchers

RAISED MORE THAN **$67 Million** for Cystinosis Research

RECEIVED **1 FDA Approved Drug, and 1 FDA Approved Clinical Trial**

*In 2022, CRF:*

AWARDED **9** **New Research Grants** in **5** Countries **Totaling More Than $1.6 Million** and PUBLISHED **6** **Articles** in Prestigious Journals by CRF Researchers

**100% OF YOUR DONATIONS DIRECTLY SUPPORT CYSTINOSIS RESEARCH**


Charlie
Simpson


# DAY OF HOPE 2023

by Stephen Jenkins, MD, Sam and Lars' dad

The 2023 Day of Hope family conference was a big success. Over 260 people from eight countries gathered in Newport Beach, California at the VEA resort to make connections, learn about the latest research, and find inspiration and rekindled hope. It was great to see old friends and make new ones as our cystinosis family continually expands.


Nancy Stack started our conference on Friday morning with an update on CRF. Since its founding in 2003, CRF has raised $67 million for cystinosis research and funded over 223 studies, resulting in over 103 publications. CRF funds research in critical areas, including gene therapy, muscle and bone disease and potential new therapies.

Dr. Julian Midgley from Alberta, Canada, shared reflections and outcomes from his 30 years as a pediatric nephrologist caring for people with cystinosis. He has seen 23 patients with cystinosis, which is an astonishing number for such a rare disease. He has become a world expert, and it is obvious he cares deeply about his patients.

After Dr. Midgley, we heard from Dr. Francesco Emma, who came all the way from Rome, Italy, from the Bambino Gesù Hospital, to share an update on the many research studies he has done over the years. Dr. Emma performed a drug-library screen for medications that could both deplete cystine and prevent apoptosis (cell death). They identified an FDA-approved medication called disulfiram. His team was excited about this discovery because disulfiram has a longer half-life and fewer side effects, and it is much cheaper than cysteamine. Unfortunately, when they tested the medication in animals with cystinosis, it was extremely toxic, even lethal. He used this example to illustrate why it is important to not take shortcuts in medical research. Just because something looks promising in initial experiments does not mean we should start giving it to humans.

He followed this up with a couple other potential compounds his team has identified. One of them helps get cystine out of cells through an alternative pathway. The other compound improved autophagy and led to less cell death in kidney cells. Both compounds require additional studies in animals before they can be tested in humans. Because mice are small and difficult to evaluate with blood tests over a long period, Dr. Emma's team developed a rat model with cystinosis so that they can perform these types of experiments.

Lastly, Dr. Emma shared some surprising data on the ketogenic diet. Mice with cystinosis treated with a ketogenic diet experienced improvement in their Fanconi syndrome. The same was true in rats. He is not advocating patients start the ketogenic diet at this time, as there may be other unexpected effects. He thinks we might be able to use medium chain triglycerides, like commercially available triheptanoin, as an alternative to the ketogenic diet. When given to animals, they still had improvement in Fanconi syndrome (although not as dramatic as when given a ketogenic diet).

After Dr. Emma, we heard from Dr. Paul Grimm from Stanford. Dr. Grimm started his talk with a discussion of the trauma that parents and people with cystinosis have experienced. He talked about the time toxicity of trying to give our children and ourselves "perfect" care. He proposed the standard of "good enough" care and emphasized that we shouldn't beat ourselves up when adherence to medications and treatments isn't 100% perfect. This really resonated with me, as I think about all the guilt I experienced after my son, Samuel, was diagnosed and we adapted to our new lives.

Dr. Grimm also talked about medication management in cystinosis. Indomethacin, widely used in Europe, is now being used more commonly in the United States for polyuria and electrolytes. He talked about the unconventional use of diuretics like amiloride to increase potassium levels, and thiazides to reduce urine output.

He shared his opinion that you don't need to rush to start cysteamine eye drops until a child is school age, because it's more important to focus on nutrition and electrolytes in the early years. He also said not to worry about giving eye drops at school, and you can give them more frequently than every hour when they get home to make up for it.

He raised a lot of eyebrows when he suggested that for some people, Procysbi® could be given three times a day instead of twice a day. If a person is taking 12 (75 mg) capsules every 12 hours, an alternative approach would be to take 8 capsules every 8 hours. You still get a full night sleep, and peak drug levels wouldn't be so high, which may help side effects and odor.

Next, Dr. Sergio Catz from The Scripps Research Institute talked about his research on inflammation in cystinosis. White blood cells called neutrophils release inflammatory proteins called cytokines and proteases in animals with cystinosis. This inflammation leads to damage and fibrosis. Cysteamine does not prevent neutrophil activation. He has discovered some compounds that might reduce neutrophil activation and prevent organ damage.

Dr. Clay Emerson, new CRF Board member and father of Brooke Emerson, gave a talk on the Cure Cystinosis International Registry, which was launched

# DAY OF HOPE 2023

in the spring of 2021. This is a new registry with updated questions that reflect the progress and changes in management for cystinosis. Because cystinosis is so rare, it is critical that as many people with cystinosis participate in the registry as possible. The patient registry is a key link between patients and researchers and will guide research towards improved treatment for cystinosis.

Dr. Benjamin "Beno" Freedman from the University of Washington gave an update on his research on induced pluripotent stem cells. His lab is able to take urinary cells and reprogram them to be stem cells, which can then form new tiny pieces of primitive kidney tissue called organoids. He has collected lots of urine from people with cystinosis in the past and created cystinosis organoids that respond to cysteamine treatment. Using CRISPR, powerful gene editing technology, he can insert the cystinosin gene into these cells, which increases cystinosin protein production, and reduces cystine accumulation. The goal is to implant these genetically modified organoids into native kidneys, where they can engraft and contribute to improved kidney function.

Dr. Morgan DiLeo from the University of Pittsburgh gave us an update on cysteamine solidrops, a new treatment for corneal cystinosis. Solidrops are a controlled-release formulation that use a gel-based eye drop that contains cysteamine-loaded microspheres. The hope is that it could be given once a day and would slowly release cysteamine to the cornea. It has been tested on rabbits and cystinosis knockout mice, and soon Dr. DiLeo hopes to talk to the FDA about clinical trials.

Dr. Reza Seyedsadjadi from Massachusetts General Hospital gave us an update on his research of myopathy and dysphagia in adults with cystinosis. His group previously evaluated 20 patients with a battery of tests including video fluoroscopy of swallowing. They were able to bring eight patients back three years later to repeat clinical testing. Performance was worse in some measurements, like Timed Up and Go and grip dynamometry, but some patients actually showed improvement in swallowing, especially related to the oral transport phase of swallowing. Dr. Seyedsadjadi did muscle biopsies in three patients and was able to isolate satellite cells which retain regenerative capacity.

In addition to hearing from researchers, we had multiple families share their experiences with cystinosis. Vanessa Bonneau and Sylvan Lanken from Montreal, Quebec, talked about their daughter Fera, who is two years old, and what it was like navigating her sickness and diagnosis as the world shutdown due to COVID-19. Despite the sadness of a devastating diagnosis, and the isolation of pandemic lockdown, they shared their story with humor and hope.

Crystal Walker and her thirteen-year-old daughter Aliyah from Calgary, Alberta, shared their journey with cystinosis and another severe rare disease, moyamoya, which has affected both Aliyah and her sister Madelyn, who has suffered multiple strokes, leaving her hemiplegic and nonverbal. Both sisters have required brain surgeries. We were all inspired by their bravery and resilience, especially as Aliyah spoke candidly about her challenges, hopes and dreams. It's hard enough to deal with one

rare disease, but the Walkers have shown that you can build a meaningful and joyful mosaic with the puzzle pieces you are given. I think that idea resonated strongly with everyone in the room.

The final talk on Friday was from Dr. Stéphanie Cherqui of UC San Diego. She gave an update on the gene-modified autologous stem cell transplant Phase I/II trial that started in 2019. She has transplanted six adults. All of them remain off cysteamine therapy and there have been no adverse events related to the drug product. She has seen a reduction in white blood cell cystine, cystine crystals in skin and rectal biopsies, and crystals in the cornea. After Dr. Cherqui's talk, we had a panel with three of the participants: Jordan Janz, Natalie Stack and Kurt Gillenberg. They took the stage like the rockstars they have become for our community. They spoke openly about the difficulties involved in getting the transplant, especially dealing with the side effects of chemotherapy, but none of them regret doing the trial. It was an amazing culmination of 20 years of research funded by CRF.

On Saturday morning we heard from Dr. Grimm again, this time talking about chronic kidney disease and kidney transplants. He talked about how children can be listed for transplant before their GFR drops below 20, which helps you accumulate wait time for when you really need it. He emphasized that a living donor kidney transplant lasts around 19 years, compared to 11-12 years for a deceased donor. He shared tips on how to help your transplanted kidney last as long as possible, including following a mostly vegetarian diet, exercising, controlling cholesterol, not smoking, and controlling blood pressure.

Dr. Emma gave another talk about long term outcomes in a large patient cohort from Europe, including 453 patients. The study showed that the earlier you start cysteamine, the longer your native kidneys will last. The average age for dialysis or transplant if started on cysteamine in the first year was 22. If started in the second year, it's 18. If started in the third year, it's 14. They also found that specific mutations, like the 57KB deletion, did not predict severity of disease. They found no difference in

late complications like muscle wasting and thyroid disease as long as patients started cysteamine before age five. They looked at patients who took indomethacin, and it was not associated with better or worse outcomes. They found the same for ACE inhibitors and angiotensin receptor blockers (ARBs). He recommended kids receive indomethacin early on to help with polyuria and electrolytes, but then stop them around the time they go to school or if starting an ACE inhibitor or ARB.

After Dr. Emma, we heard from Kathleen Roberts, a young adult from British Columbia with cystinosis. Kathleen talked about how so much of your life is out of your control, but it is important to not let cystinosis prevent you from having life experiences like any other person. She talked about going to nursing school and working as a NICU nurse, and her experience of getting a kidney transplant. She talked about the empowering effects of empathy. It was a powerful and moving talk.

Dr. Ana Francisco from Albert Einstein School of Medicine in New York City shared her research on executive function in cystinosis. My boys participated in this study last summer, so it was cool to see the preliminary results. People with cystinosis were hooked up to EEG monitors and given different tasks to perform. The EEG readouts were compared to normal controls. Dr. Francisco found, on average, people with cystinosis had no significant differences in conflict monitoring, memory updating, set shifting or response inhibition, but there was a difference in error monitoring. Participants were aware they had made mistakes but seemed to process it more slowly. This research could lead to adaptive strategies to improve executive function.

After Dr. Francisco's talk, we had breakout sessions based on ages: parents of newborn-eight years, parents of children nine-17 years, adults with cystinosis and their partners, parents of adults, teenagers with cystinosis, and a new group, grandparents of people with cystinosis. This was one of my favorite parts of the whole conference. It was a really wonderful group therapy session. It was so great to talk through things and get advice from people who actually understand what you and your family are going through. I think we could've talked for several hours.

After the breakout sessions we had a panel of adults living with cystinosis. It was motivating to hear them talk about the challenges and successes of a life with cystinosis. We heard from newly minted adults like the Partington twins and old timers like Mack Maxwell. They shared great advice on things like medication adherence, friends, school, work, and kidney transplants. I was glad my son, Samuel, was able to sit in on that session to hear from these giants.

After the adult panel we had our conference wrap-up. We shared what we had learned in the conference and what we were grateful for as a group. The words "resilience" and "hope" and "community" came up again and again, and I think that's what sums up CRF's mission. We are already looking forward to next year.

# Natalie's Wish '23

CELEBRATING TWENTY YEARS *of* HOPE

## $1,140,100 Raised
FOR CYSTINOSIS RESEARCH!

Thank you for joining us in April for the Natalie's Wish Fundraiser. Together we raised a remarkable $1,140,100 for cystinosis research. Your donations will help fund current research studies and new research studies initiated this year. With your support and partnership, we will fund novel research ideas aimed at developing new treatments and a cure for cystinosis.

It is because of you that we have reached milestones once unimaginable; a new FDA medication, an FDA approved clinical trial to test a one-time treatment that could cure cystinosis, and numerous on-going clinical trials that we hope will result in new, improved medications and treatments.

CRF is the largest fund provider of cystinosis research grants in the world. Since 2003, CRF has funded 223 research grants, eight extension grants, and nine equipment grants in 12 countries. We have created a synergistic research community dedicated to our children and adults with cystinosis. Our research teams have made discoveries that have led to significant advances in the treatment of cystinosis, bringing renewed hope to our children and adults with cystinosis that a cure is within sight.

We are grateful to you for your continuing support of our research efforts and for your commitment to Natalie's Wish!

# FAMILY FUNDRAISING from around the world

**In 2022, families from our global community contributed $1,635,355 for cystinosis research!**

YOUR GENEROSITY CONTINUES TO GIVE US HOPE — A HOPE THAT UNITES US.

## UNITED STATES

HADLEY ALEXANDER - $42,125
ISAAC ANDREWS - $25,000
ETHAN BARTKOWSKI - $200
LILY BEAUREGARD - $9,525
NICHOLAS BECHT - $300
JACKSON BLUM-LANG - $101,000
NOAH BROWN - $550
CHASE CHODAKOWSKY - $660
JOSHUA CLARKE - $10,100
MIA COPELAND - $400
BAILEY DEDIO - $510
BROOKE EMERSON - $31,000
JAMES 'DREW' ENDSLEY - $200
TINA FLERCHINGER - $41,095
COLLINS GALLOWAY - $104,434
FIDELITY GOODRIDGE - $190
CALEB GOWAN - $1,613
HOLT GRIER - $2,540
CARTER JACKSON HALL - $925
NICOLE HALL - $8,421
ANNE BARBER HALLOCK - $1,600
ANNALIESE HAMMER - $1,000
LANDON HARTZ - $38,502
JAKOBI HORTON - $200
KORA HOUTZ - $180
DIEGO HUSKER CANO - $300
SAM & LARS JENKINS - $3,100
TYLER JOYNT - $450
JOSIE KANUPKE - $8,398
SHANNON KEIZER - $250
DEVLIN KEON - $465
AARAV KHALASI - $22,292
HAYDEN KIRCHHOF - $36,978
DILLON MARIE KISH - $125
JAKE KRAHE - $64,630
AMANDA KUEPFER - $5,491
KENZIE LAWATSCH - $1,575
KALEB LAWSHE - $2,075
GENESIS LOPEZ - $125
LOLA LONG - $2,490
AYLA & OTTO MAHER - $111,315
KEEGAN MANZ - $2,450
ISLA BROOKE MCALLISTER - $306
ALEX AND MICHAL MCDOWELL - $100
STELLA GRACE MILLER - $3,810
BRADY MURDOCH - $4,024
AIDAN O'LEARY - $33,125
JENNA AND PATRICK PARTINGTON - $157,850
EMMA AND GRACIE PATTERSON - $1,325
KELLY PFEIFER - $202
ABEL & PAUL PRUITT - $55,020
ZACH AND FRANK RITCHIE - $200
CAMDEN SANDERS - $110
EZEKIEL SHIELDS - $175
CHARLIE SIMPSON - $364,297
ZYLAR SMETHURST - $200
BRIAN SMITH - $500
CIENNA SMITH - $2,000
MITCHELL SMITH - $650
SOFIE SOS-FINUCANE - $32,001
GABE STEPHENSON - $250
HENRY STURGIS - $125,255
EMMA GRACE SUETTA - $6,648
PEYTAN TAYLOR - $100
LEANNE TREMBLAY - $293
BRADEN AND DAX TYNER - $750
ALEX WEAVER - $300


## CANADA

SOPHIE'S CHAMPIONS
SOPHIE BETOURNAY - $27,250

FERA LANKEN BONNEAU - $500

NORA & ALAN CAMPBELL - $273

SETH'S CIRCLE OF HOPE
SETH deBRUYN - $25,154

HOPE FOR JAMES
JAMES FEHR - $20,231

NOLAN LAFERRIERE - $165

MARVELED BY MADDIE
MADDIE LAWRENCE - $1,082

LIV-A-LITTLE FOUNDATION
OLIVIA LITTLE - $2,349

CILLIAN MCQUILLAN - $822

KATHLEEN ROBERTS - $2,200

GABBIE'S WISH
GABRIELLE STRAUSS - $1,795

MADELYN & ALIYAH WALKER - $950

## AUSTRALIA

ETHAN FENN - $590

BEE ROBERTSON - $882

## ENGLAND

JULIETTE FOLLAIN - $12,050

THE HUTTON SISTERS - $104

## FRANCE

AUGUST FERBER - $222

## ITALY

VIOLA PAPPACENA - $145

## MALAYSIA

LEE ZHUN HOONG - $264

## NORWAY

DENIS' FIGHT FOR A CURE
DENIS LILLAND - $537

## SLOVENIA

AJDA GRUDEN - $200

## SWEDEN

KAROLIS SCHROËDER - $212

## IN MEMORY

EDWARD, SUSAN AND JOSEPH BEAMS - $600

TANNER EDWARDS - $104,885

KIMBRA CHEYENNE GIBBS - $325

SUSAN ELLYN THOMAS - $1,100

THE HELBLING CHILDREN - $200

JESSICA JONDLE - $3,100

SARAH MELANG - $1,000

ALICE AND TRACY STRICKLAND - $250

DEBRA GEHRKE TOWERY - $1,625

PRESTON TOWRISS - $500

WESTON TSCHANNEN - $5,000

KURT WAGNER - $400

## FROM FACEBOOK

FACEBOOK FAMILY AND FRIENDS FUNDRAISING EVENTS AND DONATIONS ARE INCLUDED IN THE TOTALS ABOVE.

## GIVING TUESDAY SURPASSES GOAL – $328,741 RAISED FOR RESEARCH


Your generosity on Giving Tuesday was astounding! Together, we raised **more than $328,741** for cystinosis research! One hundred percent of the donations will go directly to research that is focused on finding novel treatments and a cure for cystinosis.

As a result of your commitment to CRF's research program, CRF has awarded 223 research grants in 12 countries, ensuring that there is a CRF researcher working on cystinosis every minute of the day. You have been the driving force behind every CRF achievement, every milestone and every step forward. Your dedication to our children and adults with cystinosis made our hope for a cure possible. Thank you for being part of the Cystinosis Research Foundation community.

## SINCE 2016, CANADIAN FAMILIES HAVE FUNDED $1,011,762 FOR RESEARCH IN PARTNERSHIP WITH CRF

**Canadian cystinosis families are committed CRF partners working with CRF to fund research that will lead to better treatments and a cure. Families across Canada continue to organize and plan events to raise money for research. In 2022, Canadian families raised $108,646.73 (CAD) for cystinosis research.**

The Cystinosis Awareness and Research Effort (CARE) has partnered with Canada Helps to establish the Canadian Cystinosis Research Foundation. This fund is administered by Aqueduct Foundation and allows for an efficient and effective fundraising process, ensuring that Canadians who donate will receive a charitable tax receipt. Canadians also have the option to donate directly to CRF if they so desire, however, no charitable tax receipt will be issued.

When it comes to finding a cure, there are no country borders! Working together, our two countries have united efforts to raise awareness about cystinosis, to advocate on behalf of all children and adults with cystinosis and to ensure that we will fund the most qualified researchers in the world.

Since 2016, Canadian families have funded $1,011,762 in grant installments for CRF research studies through the Aqueduct Foundation. CRF is grateful for the support of our Canadian cystinosis families and friends who work tirelessly to find better treatments and a cure for cystinosis.

Canadian families are creative in their organization of fundraising events, supporting CRF research through everything from lemonade stands to bake sales to golf tournaments, building their sense of advocacy and community engagement and having fun along the way.

If you are Canadian and want to donate or raise money for CRF sponsored research, contact one of our CRF Board Members for information. Barbara Kulyk at *barbiekulyk@hotmail.com* or Kristen Murray at *murraykristen@hotmail.com*.

# KRISTEN MURRAY

Together with my husband, Nathan deBruyn, and our 11-year-old sons, fraternal twins Seth and Leif deBruyn, I live in Calgary, Alberta, in the foothills of the beautiful Rocky Mountains. As a family, we strive to live a healthy, active lifestyle, enjoying activities like soccer, gardening, hiking, camping, mountain biking, and canoeing, close to home and beyond.

Before starting my family, highlights of my life included trekking in the Himalayan mountains of Nepal, studying French in France, rock climbing in Thailand, cycle-touring in Iceland, canoeing the remote Wind River in northern Yukon and marrying Nathan in a rustic winter wedding complete with down jackets and cross-country skis.

In welcoming Leif and Seth into the world, we would embark upon the greatest and most remarkable adventure of our lives. Seth's diagnosis with cystinosis at 18 months certainly created deep shadows on our path. I now marvel at the light that began to shine soon after Seth's diagnosis when we first learned of CRF. Since those very early days, CRF has continually shone rays of hope on our path. Our family connection with CRF has had a profound influence on our lives.

Over the years, I have derived strength and joy from my contributions, however small, to CRF, regarding them as opportunities to make meaningful connections and to infuse our journey with cystinosis with purpose and hope. In October 2014, "Seth's Circle of Hope" came to life as Nathan and I reclaimed the once ominous date of Seth's diagnosis. It has since become an annual event in which we join with friends and family, lighting candles and supporting CRF in Seth's honour.

In addition to fundraising, I have taken part in several CRF Day of Hope Family Conferences, submitted writing to *Cystinosis Magazine* and enlisted Seth in research projects. Most recently, I collaborated with the organizing committee for the Cystinosis Research Foundation Pre-Session Conference as part of the International Pediatric Nephrology Congress hosted in Calgary in September 2023. In addition to assisting with session planning and an after-event dinner, I was thankful to share the story of my family's journey with cystinosis as a presenter.

I hold a bachelor's degree in political studies and psychology, an education degree and a Master of Counseling. Beginning my teaching career abroad in South Korea and Turkey, I combined a love of travel and adventure with what would become a passion for developing supportive relationships in which students could learn and grow. Over the years I have been honored to teach English as Another Language to adult and youth English language newcomers. I have also earned my certification as yoga instructor and have taught yoga, mindfulness and stress reduction to high school students. In my current role as a high school guidance counselor, I support vulnerable populations and help the young people with whom I work to see and take care of their best possible selves.

In my roles as teacher, counsellor, mother and member of the cystinosis community, I try to embrace all that life has to offer, including, and perhaps especially, the challenges that come along. I endeavor to find strength and purpose and to harness individuality and creativity in the face of adversity. I strive to instill meaning and hope, to cultivate positive change through thoughtful and dedicated action in the myriad of pathways of my life, and to encourage others to do so too.

Margaret Mead said that we should "never doubt that a small group of thoughtful, committed citizens can change the world." In its 20 years, CRF has changed the world. Blazing new trails of research, bringing forth better treatments, moving ever nearer to a cure, CRF has brought relief and real hope to so many. I am excited to serve as a member of the CRF Board of Trustees. I look forward to this opportunity to work alongside others who are committed to CRF and to do all that I can to support the organization and the community that mean so much to my family and me.

Read more about Seth's Circle of Hope candle lighting on page 53.

# CLAY EMERSON, PhD, PE, CFM

Our daughter Brooke is a beautiful, thoughtful and imaginative eight-year-old girl. Our family's journey with cystinosis began with Brooke's diagnosis at 16 months. Since her diagnosis, Jill and I have done everything in our power to keep Brooke healthy and happy. After learning of her diagnosis, we immediately became aware of the research that the CRF was funding and the impact it was having. We were in contact with other CRF families who provided an informed ear to listen to us and graciously gave us advice and much-needed comfort.

As an engineer and scientist, I understand the value of research. As a father of a child with a rare disease, I know that research is of paramount importance. Although Brooke's diagnosis was a difficult time for our family, I found hope in knowing that there was an organization that was focused on research for better treatment and a cure for cystinosis. Just a couple months later we were on our way to our first Day of Hope event. The conference always reminds us that we are part of a community and that there is hope for better treatment and a cure.

As Brooke's father, I want Brooke to lead a happy, healthy and full life. And while cystinosis presents many daily obstacles, Brooke has already benefited from CRF-funded research efforts. Since Brooke's diagnosis, each year we've had a CRF fundraiser. So many of our friends and family have graciously participated, helping raise money for ongoing research efforts. This year we had our eighth successful fishing fundraiser (www.hopeforbrooke.com).

I am honored to have been nominated for the CRF Board of Trustees and believe I have the necessary personal and professional experience to be a contributing member. I have a BS in civil engineering from Rowan University, an MS in environmental engineering from Drexel University and a PhD in engineering from Villanova University. I am a licensed Professional Engineer and a Certified Floodplain Manager. I have worked at Princeton Hydro, a consulting engineering firm, for the past 15 years. I am also an adjunct professor at Villanova University where I teach a water resource-related course in the College of Engineering.

In summary, my goal is to do anything in my power to help Brooke and other people with cystinosis. I believe that advancing the mission of the CRF is the way to accomplish this goal. I am honored to work with the CRF Board of Trustees.

Read more about Hope For Brooke's fishing fundraiser on page 52.

"Never doubt that a small group of thoughtful, committed citizens can change the world."
–Margaret Mead

It Takes A Village

# CYSTINOSIS COMMUNITY CALENDAR OF EVENTS

We would like to acknowledge all families for their support of cystinosis research, unfortunately some events may have passed by the time this issue is mailed.

## Friday, June 2, 2023

### LOTS OF LOVE FOR LANDON CHARITY GOLF OUTING
IN HONOR OF LANDON HARTZ
Black Hawk Golf Course, Beaver Falls, Pennsylvania
*Contact Jimmy Hartz: lotsofloveforlandonCRF@gmail.com*

## Saturday, June 3, 2023

### LILLYANNA'S LEMONADE FOR A CURE
IN HONOR OF EMMA GRACE, LILLYANNA'S SISTER
440 Pig Alley, Etna, California
*Contact Shelly Suetta: shellysuetta@hotmail.com*

## Sunday, June 25, 2023

### FOURTH ANNUAL CONCERT FUNDRAISER
IN HONOR OF LILY BEAUREGARD
East Warren Rod & Gun Club, Warren, Rhode Island
*Contact Tom Wallis: thomaswallis@cox.net*

## Thursday, September 7 - Friday, September 8, 2023

### CRF INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM *(BY INVITATION ONLY)*
Mabel and Arnold Beckman Center, Irvine, California
*Contact Nancy Stack: nstack@cystinosisresearch.org*

## Saturday. September 16, 2023

### AARAV'S TIME TO SHINE GRAND GALA
IN HONOR OF AARAV KHALASI
White Lotus Banquet Hall, Citrus Heights, California
*Contact: info@aaravstimetoshine.org*
*or Visit: www.aaravstimetoshine.org/grand-gala-2023*

## Fall 2023

### CAPITAL CUP GOLF TOURNAMENT
IN HONOR OF JENNA & PATRICK'S FOUNDATION OF HOPE
Sacramento, California
*Contact Kevin Partington: kevin.partington@cushwake.com*

## October 2023

### FIFTH ANNUAL CHILI COOKOFF FOR CYSTINOSIS RESEARCH
IN HONOR OF LILY BEAUREGARD
Eagle Event Center, Fall River, Massachusetts
*Contact Shelli: shellipereira@gmail.com*

## Saturday, October 21, 2023

### SETH'S CIRCLE OF HOPE
IN HONOR OF SETH DEBRUYN
Calgary, Alberta, Canada
*Contact Kristen Murray: murraykristen@hotmail.com*

## Tuesday, November 28, 2023

### GIVING TUESDAY – FUND A CURE FOR CYSTINOSIS!
CYSTINOSIS RESEARCH FOUNDATION
*Visit: www.cystinosisresearch.org*

## February 28, 2024

### RARE DISEASE DAY
CYSTINOSIS RESEARCH FOUNDATION
*Visit: www.cystinosisresearch.org*

## Month of April 2024

### NATALIE'S WISH CELEBRATION FUNDRAISER
CYSTINOSIS RESEARCH FOUNDATION
*Contact Nancy Stack: nstack@cystinosisresearch.org*

## April 2024

### DAY OF HOPE FAMILY CONFERENCE
CYSTINOSIS RESEARCH FOUNDATION
Location: TBD
*Contact Nancy Stack: nstack@cystinosisresearch.org*

20 years TOGETHER

# FAMILY STORIES

# CYSTINOSIS HAS GIVEN US PURPOSE

By Asim Bukhari, Rayan's father
DHAHRAN, SAUDI ARABIA

Almighty Allah does not burden a soul but to the extent of their ability. I will start from where I left off last time. I verily believe we are not given a pain that we cannot bear — we are not given a goal that we cannot achieve.

We, humans, are basically very emotional people, individually, socially, religiously or politically. We should always believe that in every problem there is a solution and if we try our best, success is our destiny. Face the difficulties — because there is no difficulty in your life that no one else has faced. There is no tragedy that does not happen to anyone else. And of course, there is no success that no one else has.

Being a Muslim, my faith in religion is quite natural, especially in the biggest test of my life. With every hardship comes ease and this has been seen in the past years of my life. Too much knowledge is painful too, but knowledge about cystinosis has certainly made our life easier than before. Now we enjoy our son Rayan's mischiefs and talking more. My daughters, who used to worry about the physical condition of Rayan while playing with him, can now play with smiles and laughter. Our family life is as if you are walking on stones and a river flowing underneath and there is beauty on both sides. Now, on one hand, you have to save yourself from falling into the water and getting hurt, and on other hand need to enjoy the scenery. In any way, it is, life is better now and it will be better in the future, God willing.

My main focus of writing today is all on parents, friends and society. The biggest problem of our society is aimlessness; in our childhood, our life had no purpose, we grow up, we become young and we become old but, in our life, no goal. We spend our childhood and youth in education, this education gives us a degree and we also get a job on the basis of this degree, but again, this degree and this education also do not make our life meaningful. You must have seen this with many people around you. This meaninglessness is actually a lack of purpose.

In our life, aimlessness often increases and we become sad, but these special children bring us out of this sadness and save us from uncertainty. These special kids are healthy, beautiful and smart and they know how to move forward in life. And through these special children, Allah has also given us the purpose of life. The path and the journey might be difficult but the destination is not far and this peace turns into happiness and shines in our faces, in our eyes.

Another such goal has also been given to us by the Cystinosis Research Foundation. It has given us the courage to see life from a different angle, from which it looks even more beautiful and I salute all the scientists, doctors, management and cystinosis community for making life more meaningful, and more thankful. True, life never stops. One world and many stories…

THROUGH THESE CHILDREN, ALLAH HAS GIVEN US THE PURPOSE OF LIFE.

*The path and the journey might be difficult but the destination is not far and this peace turns into happiness and shines in our faces, in our eyes.*

## FISHING FROM SUN-UP TO SUNDOWN

See Brooke's catch on page 52

HOPE FOR BROOKE

FISHING FOR BROOKE'S CURE

# A Small but Mighty Fundraiser

By Jill Emerson, Brooke's mom
HAMMONTON, NEW JERSEY

We had our first Fishing for Brooke's Cure fundraiser in April 2016, less than two months after Brooke's diagnosis. While in some ways we felt paralyzed and overwhelmed with the prognosis, we felt we had no other option but to begin fundraising immediately. We did it out of necessity – necessity to do something about our daughter's future during a time in which we felt powerless, a necessity to pay it forward and give back to the cystinosis community and people who had been fighting this fight for years before ours began, and a necessity to keep ourselves busy so that we didn't let that initial dread consume us.

It started out small (and it still is!), and we came up with a fundraising idea that had little to no overhead costs, took minimal planning time and effort, and focused on something already a part of our everyday lives – fishing! Clay, who is an avid fisherman, invited his four friends on a fishing trip. Nothing that isn't an ordinary occurrence on a spring day. The catch? They would also be raising funds for Cystinosis Research Foundation. Each fisherman sent a simple email to their family, friends, and colleagues, explaining the rules and encouraging them to pledge a dollar amount per fish caught. And the first annual Fishing for Brooke's Cure fundraiser was born.

It's become a competitive event, with the fishermen fishing from sun-up to sundown one day each Spring to catch the most fish. It's also a bonding experience for them, as they get together and craft lures before the event and have turned the event into a two-night trip down south. The competitive but friendly banter between the fishermen begins earlier and earlier each year. Even during the pandemic, we were able to safely have our fundraiser, and it brought a lot of connection and happiness to an otherwise isolated time.

We've tried different things throughout the years in an effort to raise more funds, but I won't lie. There have been times where we get discouraged, wishing we had a larger community to engage and could raise more funds, and try to creatively think of additional fundraising opportunities and events we could have.

But while we don't have a big community, we have a loyal one. We aren't raising large amounts of money each fundraiser, but over these past seven years, we have raised over $222,728 through our grassroots fundraiser. Through our small but committed donors, we have raised enough money to fund a 2-year research study! When we think about it from that perspective and realize the impact our tiny fishing fundraiser has had on funding cystinosis research, it reminds us that no donation is too small, no fundraising effort not worth it, and even small but mighty communities can have a profound impact on the lives of those with cystinosis.

We are eternally grateful to our fishermen who are committed each year to reaching out to their family, friends, and co-workers, to each donor who has donated even $1 on Brooke's behalf, and to the Cystinosis Research Foundation for always cheering us on, and for helping us recognize that our efforts can make a huge difference in the lives of others.

The past few years have been very busy, as I graduated high school and am currently enrolled in college. Last year I had planned to go to University of Idaho, 45 minutes away from home, close but far enough away. I was so ready to be on my own and wanted to join my friends as they all were headed up there. Fast forward to now, I am attending a local state college and living at home.

# Shifting Energy

Last year, when I was a senior in high school, I got COVID-19. I am not really afraid of getting sick; it is just annoying as it knocks me down more than the average person, because of cystinosis and my kidney transplant.

After COVID-19 I wasn't feeling right; my kidney felt swollen and I had more aches than usual. One day after my blood draw, I found out my kidney was rejecting. I traveled to the hospital 350 miles away and was put on the heaviest drugs with the highest doses. The drugs were so strong my veins gave out on me, and I went through five IVs in four days. I was so big and puffy because of all the steroids, and so down on life. The fact that my kidney was rejecting didn't make any sense, as I took care of myself well.

In January and three kidney biopsies later, I was out of rejection. After many medical challenges, I graduated, and a few days after graduation, I was in the hospital for my double nephrectomy surgery. This surgery was to remove my old kidneys because they were still functioning at a low level, causing electrolyte loss, dehydration, fatigue and constant urine output. A few months later when I started college in August, I was feeling better. Sure, I had to go to the emergency room that summer, but it was nothing new. My kidney was finally working.

Fast forward to now, my kidney is stable. It is at an extremely low functioning rate, but I feel good, which is all that matters. I am getting ready to move out this fall and attend the University of Idaho. I am majoring in psychology right now with the goal of becoming a clinical pediatric psychologist. I want to help kids in hospitals with their mental health. I had a doctor who listened to me when I was in the hospital, and it really made a difference in my attitude and determination to not give up.

Nowadays, I am attending college, working, swing dancing and going to the gym. I like to stay active and push myself when I am feeling good. I honestly push myself constantly, even if I have no energy. I push myself to go to the gym, run a mile and lift weights. It can be a good thing, but I can overdo it. So, I have been working on balance. I do try and see life from a positive perspective.

One saying that always gets me by is the thought that, "It could always be worse." This, and trying to rely on God, are what gets me by. All in all, I am living my life and making something of my challenges in the hopes that someday I can help others like me through their challenges in life.

By Tina Flerchinger
CLARKSTON, WASHINGTON

LUCAS HEITZ

# Full of Hope

Our little boy Lucas is two years old. When he was seven months old, he stopped growing and gaining weight. After many examinations, including genetic testing, the diagnosis of cystinosis was made.

We learned of his illness on May 10, 2022, so it's only been a few months since we've learned to live with daily medications and regular medical appointments.

It was a shock for us because we didn't know what was happening to us despite the fact that I had done a lot of research beforehand to find out what our son could possibly have. Among all the things I had researched, there was cystinosis, so the day our doctor announced the results, I already knew what we were going to face, but only in theory.

We went through a lot of different feelings, including anger, guilt and sadness. For us, the main thing we focused on was staying strong for Lucas. We wanted to continue to live normally for him but also for his 8-year-old brother. We are surrounded by our loved ones, our family and friends. They support us, listen to us and help us if needed.

It took us a few weeks of taking the medications before it became a habit and fit into our daily lives. We preferred to change some of our habits so that we did not need to change Lucas' pace too much. For example, we eat earlier or later depending on the time that Lucas takes his medications. We put his supplements in his bottle of water since he drinks all day. At night, he has one bottle of water in his bed along with a night light, so that he can drink if he needs to.

He also takes Procysbi® in the morning and evening. In the beginning we tried lots of methods to give him the medication because it was not easy to do since the small medication beads stick to everything once in contact with water. In the beginning we used a bottle nipple that we cut so that there was a larger opening at the end. We placed the small beads inside the nipple then we poured the water on it and Lucas was taking it and it was going well. Today he takes the medicine without any problem; we put the beads in a glass of water and he drinks it with a straw. We started a new drug, Indocin, a short time ago, and at the moment, he tolerates it well.

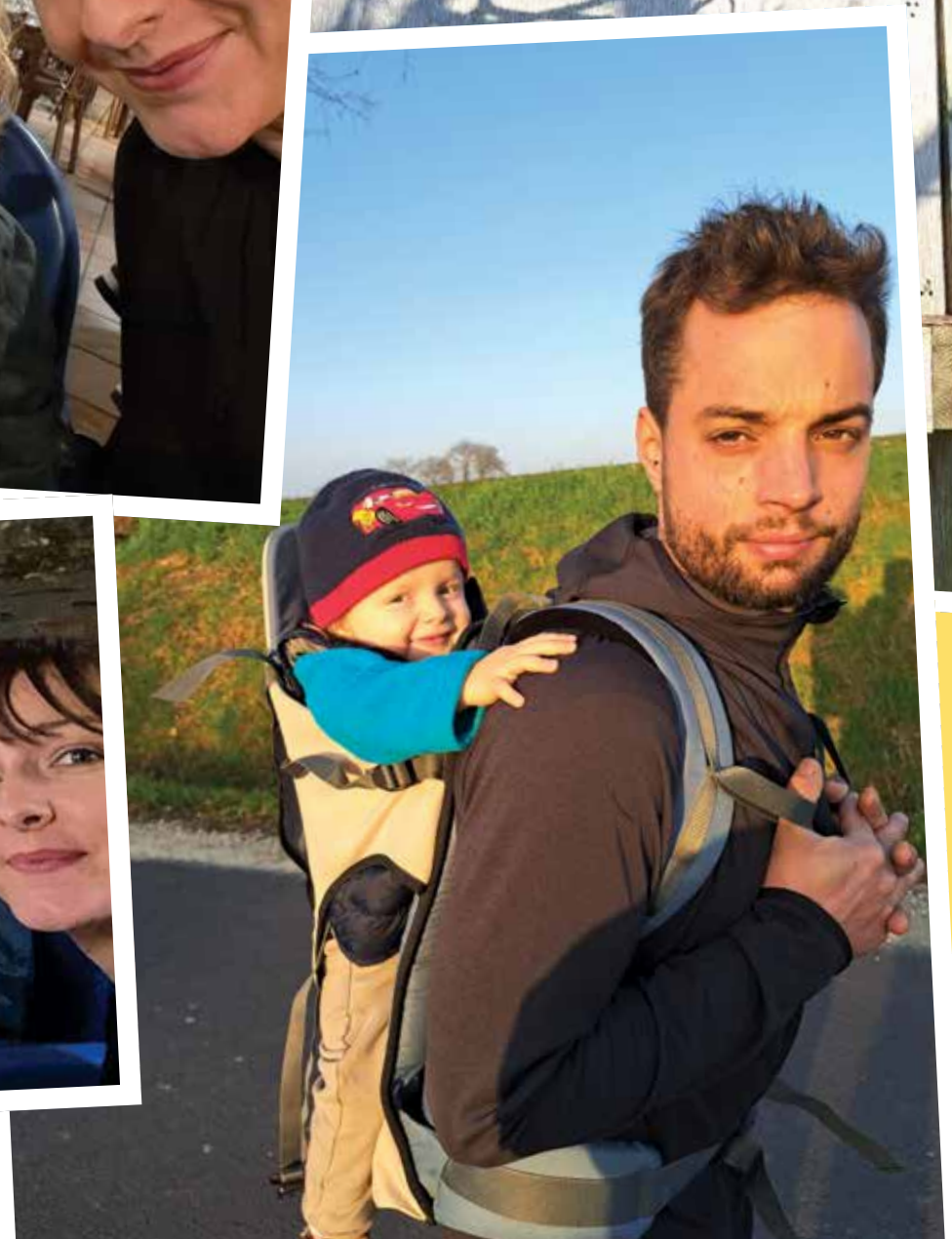
Lucas is a little boy who understands a lot of things for his age. We are lucky because he eats very well and takes all his medication by mouth because we have chosen not to have a nasogastric tube or gastrostomy either.

We had the opportunity to go to Leuven in Belgium to attend the cystinosis conference in July 2022. It gave us hope to see the researchers and doctors who are doing so much research on this disease.

**WE CAN ONLY HOPE FOR ONE THING:**
**THAT ONE DAY SCIENCE FINDS A MIRACLE CURE.**

Today Lucas has resumed his growth, and we hope that will continue. He is a little boy full of life, playful and always smiling. We send a lot of courage to all the cystinosis families who, like us, have a little hero or a little heroine who gives them strength to move forward with their smile, their strength and the love they give us. Our son, our rare pearl, Lucas.

By Laetitia Heitz, Mom to Lucas, 2 years old
BRITTANY, FRANCE




Excerpts from Caleb Gowan's speech delivered at his high school graduation in May 2022.

# Raise Up

Caleb currently lives independently and works at the local gas station/ convenience store while he decides on his future endeavors. He is well known in his home community of Karlstad, MN and through his employment and everyday living, we witness the ease in which he goes about his day sharing his zest for life with those around him. So many people have "raised up Caleb in the ways he should go and he is not departing from it." For that, and for him, we are truly grateful. As Caleb's great-aunts, we are extremely proud of him. We are proud of what he has accomplished, but more importantly, we are proud of who he is as an individual. It's his daily perseverance and determination that make him an exceptional human being. His words illustrate a glimpse of who this young man is and the fine adult he is becoming.

By Janet Nysetvold and Janel Kresl, Caleb's Great-Aunts
KARLSTAD, MINNESOTA

‘Raise up a child in the ways they should go and when they are old, they will not depart from it,’ – Proverbs 22:6. This is our school’s Bible verse. It exemplifies everything Heritage Christian School and our families attempted to do and teach.

I want to address the **faculty and staff of Heritage School** with my personal and heartfelt gratitude. Living with the chronic illness of cystinosis is difficult. Your constant support, understanding, modifications, and most of all your prayers have been some of my greatest blessings. Since my diagnosis at age four, I have surpassed the goal of living to age ten. I look forward to surpassing future goals with your continued prayers.

I do not want to make my grandmas cry, but no matter what I say they probably will. **Grandma Carol and Grandma Karen**, you are two of the most selfless people I have ever met. You have always been there for my parents, for me and for others who need you. Both of you lost your husbands, my grandpas, in recent years, yet you still believe in God’s goodness and celebrate every day. It is this example of living your faith and gratitude to God that has gotten me through some tough times in my life.

**Mom, Dad and Nadine,** thank you for always loving me and accepting me, no matter the situation I was in. I hope I have made you proud and cannot thank you enough for everything you have done for me. Through my school years, basketball, cystinosis, a kidney transplant, relationships, hardships, and happy times, you were there, and I am very grateful.

To **my siblings**, thank you for loving me and spending time with me. Know that even when you bug me, I love you. I am here for you, today and always.

To **all friends** who have gathered here, thank you for your assistance in encouraging us to become stronger individuals.

To **my classmates**, I wish each of you only the best in the future. Remember, Heritage and our families have done their part. They have raised us up in the ways we should go. And now it is up to us. We must not depart from it. Thank you.”

# LIFE OF A THIRTEEN-YEAR-OLD WITH CYSTINOSIS

By Samuel Jenkins
SALT LAKE CITY, UTAH

My name is Sam Jenkins. I am thirteen years old and I have cystinosis. I've had it since I was born, so I'm kind of an expert. You would be able to tell from my science grades. I have a brother, Lars, who also has cystinosis, and a sister, Birdie. I also have two dogs, and my dad has a tortoise.

I love most food, but my favorite is almost any type of soup. One time when I was little one of my cousins asked my mom how I could eat a McDonald's Happy Meal with a G-tube. I take over fifty pills a day and got my G-tube out in January 2022.

I like to collect "Star Wars" Legos and video games from "The Legend of Zelda" series. I like walking home from middle school with my friends and watching movies. I am currently slowly writing a book inspired by my favorite book series, Redwall.

I may have cystinosis, but it's surprising how little it affects my regular life. I was recently in my school's production of "Aladdin," and before that "Matilda." Last summer I was paid two hundred dollars to participate in a brain study for cystinosis in New York City. I spent most of the money on Lego sets.

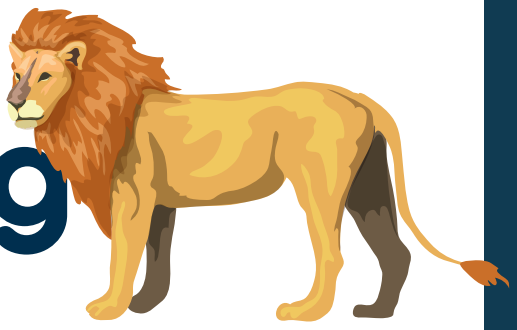
I also went to San Francisco and Monterey, California last summer, and I kayaked in Monterey Bay with sea otters. When I grow up I want to be a wildlife biologist and work with otters in Monterey. I want to get a masters degree at Brigham Young University and a PhD at Stanford. I also want to be an author as a hobby.

When I'm at the Day of Hope conference I like to hang out with my friends Henry Sturgis and Tina Flerchinger. One tradition I have with Henry is getting sushi at the Day of Hope conference every year. For me, having cystinosis is bad and unbearable only twenty percent of the time. The rest is just Spring Break fun.

My brother, Lars.

ME

# AIDAN O'LEARY
# A Burgeoning First Grader



By Erin and Jim O'Leary, Aidan's parents
CHICAGO, ILLINOIS

**It is hard to believe** that it's been six years since Aidan was diagnosed with cystinosis. Aidan just turned seven years old and is an inquisitive first grader who loves school, his younger sister Maeve and dog Gus, playing with friends, everything *Wild Kratts*, skiing and basketball. We are happy to share that Aidan remains very healthy and stable within his disease. He continues to shine his bright light upon everyone around him despite the many daily challenges he must overcome while living with cystinosis.

Aidan continues to exude bravery, strength and happiness amongst all the struggles. It's hard to think about the fact that Aidan, and all those afflicted with cystinosis, have endured more trauma and hardship than many people will ever face in their entire lifetime. While this is not the life we envisioned for him, we are awed by his grace, empathy and resilience. We used to think of cystinosis as a storm cloud that followed us around, and we were never sure when or what the storm would bring. This remains true, but we now feel a little better prepared for the storms. This year's flu and cold season has been particularly turbulent with many illnesses and double viruses, but we are doing our best to navigate the swells.

Life with cystinosis is forever evolving. The younger years were difficult with the endless trial and error until we finally found a daily medication and blend schedule that Aidan could tolerate without throwing up. We eventually were able to establish our new normal and began branching out with Aidan transitioning to school and fun extracurricular activities. We expanded our family welcoming beautiful Maeve O'Leary, and finally moved out of the city and into the suburbs.

Aidan has always had an insatiable thirst for knowledge and is very self-aware. He has always asked a million questions. He used to ask, "Why are dinosaurs extinct?" or "Why do worms come out when it rains?" Over the last couple of years, he began asking questions like "Why do I have cystinosis?", "Why doesn't Maeve or any of my friends have cystinosis?" and "If I take all of my medication, will my cystinosis go away?" These are some tough questions, especially when they are being asked by a five-year-old.

Over the last two years, we've worked hard to empower Aidan and our family with getting the support we need to better communicate and work through the challenges of living with a chronic disease. Aidan has really enjoyed play-based therapy where he can use play to find meaning; and work out life and school stressors or medical themes such as blood draws, IVs, tube changes, etc. We've also found that cranial sacral therapy has been wonderful with decreasing some of the inefficiencies within his little body.

This past year, we've seen Aidan growing into a little boy who can now better advocate for himself. We've watched as he compliantly stops a play date for meds, explains to a friend why he has a tube and needs medication, and then goes back to having fun. We've watched as he understands and helps other children that may be having a tough time. He is learning to live his life within this disease and not letting it define him. Although he has had to endure too much already, it has helped shape him into the kind, empathetic and resilient little boy he is today, and we couldn't be more proud of him.

With love,
Erin and Jim O'Leary


FIRST DAY
OF SCHOOL!

# This one's for Tucker

By Teresa Partington, Jenna and Patrick's mom
SACRAMENTO, CALIFORNIA

The Partington update memorializes our steadfast comfort animal of 12.5 years. Tucker arrived in our lives when the twins were five years old. The kids took him to show-and-tell at kindergarten, so proud of the gentle giant he was. Every day of school from K-8th grade, Tucker rode along in the car's front seat to pick up and drop off, as big as a human riding shotgun. Tucker knew how to position himself, all his weight on his hips, sometimes his front leg on the armrest, to ride along comfortably, his head out the window for a nuzzle each day as "his kids" left for school.

Tucker's death comes just two months before Jenna and Patrick graduate high school, making his life "bookends" to the kids' time growing up in our family home. Tucker saw the days when caring for two kids with cystinosis was laborious and physically exhausting for Kev and me, not to mention the kids, who were so often unwell. He saw his kids taking 1 a.m. doses of cystagon when they were on the six-hour dosing schedule. He was at our feet during countless bedding changes. He snuggled next to anyone who was home sick from school.

He was by my side during my cancer treatment. He was beside Jenna and Patrick as they recovered from the most challenging aspect of cystinosis thus far: the bone pain associated with multiple orthopedic surgeries. Tuck was with his kids in every first day of school photo, Christmas picture and snapshot on the beach in McCall.

Tucker was even a part of the Cystinosis Research Foundation's RP103 drug trial at Stanford. When the kids were six years old and in first grade, they were able to participate in the trial for the life-changing drug that is now called Procysbi®. We stayed at a Marriott Residence Inn for weeks, our loyal companion Tuck with us the whole time. He was always happy to spend time in the car with the windows cracked, but on a few long days at the study center, Tucker was allowed to come into Jenna and Patrick's research lab suite, spending the scary days of countless blood draws right beside "his kids". Tucker was an uncertified, but very capable, therapy dog. He was a big part of raising two great kids who are ready to set off into the world, cystinosis a "thing" but not the only thing by any means.

When Tucker was new to us, I would comment, "Tucker will probably live about 12 years, so when the kids are getting ready to leave for college, he will be leaving us, too." When I said it, that moment seemed so far off.

And here we are.

With Love, Teresa, Kevin, Patrick & Jenna

# Looking back on 15 years

**March 25th, 2023 marked the 15th year that the 24 Hours for Hank** foundation has put on our annual ski event fundraiser. What started out as a way to raise research dollars for better treatments and a cure for cystinosis has led to so much more. The lifelong friendships and community involvement that have been created will be everlasting.

Although our family has had a vacation home in Sandpoint since the mid '80s, in 2008 we were new residents, moving to Sandpoint only two years before. When we had our first fundraiser in September 2008 (a 24-hour bike event), we had no expectations, and honestly, were just hoping some people would show up. To our surprise, 100 people rode their bikes for 24 hours straight and raised $30,000 for cystinosis research! Henry was two at the time and rode a few miles with his training wheels. I remember him saying to me while we were riding, "See Dad, I can ride a bike. I don't need people to ride for me." Little did he know at the time what we were really doing. We made a lot of new friends that day, and that was just the beginning.

Only 4 months later in January 2009, we hosted the first 24-hour ski event at Schweitzer Mountain Resort (24 Hours of Schweitzer). I look back now and wonder how we ever did that. A few of us contrived the event while riding the 24-hour bike ride and pitched the idea to the resort in late September. For 10 years, the ski event fundraiser was followed on Saturday night with an Awards Dinner/Auction. We were basically having two events in 48 hours. Although only a small group of people did the pre-event, planning it took over 40 volunteers to run the event. Some were long-time family friends, and others just people who wanted to get involved somehow and are now great friends.

With the impact of COVID-19 in 2020, the auction moved online, and the awards dinner was no more. The ski event continued with a new format that was introduced in 2018. It was no longer a 24-hour ski event and was replaced with the longest Giant Slalom race in the United States, a 2,400 vertical foot GS course that stretched out over 2 miles and took the fast racers nearly three minutes to complete. Introducing the new format brought more new people and gave the people that had participated every year something new. It is a majestic time at the start on the top of the mountain at 7:00 a.m., watching the sunrise and listening to all of the conversations of why they are there, how many years they have been participating, or what it means to be here.

March 25th, 2023 also marked the last ski event fundraiser. It was a difficult decision to make with mixed emotions, and now that it is done it feels bittersweet. I'm sure next winter we will be wondering what to do with ourselves. It was the right decision though. After 15 years of relying on so many people and local businesses to help us, it is time to enjoy all the new friendships that we have made, in a new way. I look forward to spending time with old and new friends, reminiscing about the beautiful sunrises that we enjoyed together, or skiing in the rain, or freezing our butts off on the chairlift at 4 o'clock in the morning at past events.

I can honestly never thank the community, our family, and friends enough for all of the love and support they have shown our family and cystinosis research over the last 15 years. Without them we wouldn't be in the place that we are today. Thank you!




By Brian Sturgis, Henry's dad
SANDPOINT, IDAHO


First flyer in 2009 >


< This year's flyer

LANDON HARTZ

# PLAYBILL

## LOTS OF LOVE FOR LANDON

# Next Level Confidence

By Lauren Hartz, Landon's mom
PITTSBURGH, PENNSYLVANIA

The comfort level with being in a spotlight varies from person to person. My moments in a spotlight were few and far in between, mostly consisting of announcing that I was taking our dog for a walk on Facebook to people who ultimately didn't care, until Landon was diagnosed with cystinosis. At that time, we wanted to be noticed, because raising money for research and cystinosis became a priority. I gave speeches in front of 100-500 people, depending on the event, a couple of times a year. I wrote articles and shared pictures in the Cystinosis Research Foundation magazine and people reached out to share that they read, and were touched by, those articles. Jimmy and I chose to put ourselves in the spotlight, and by default, we put Landon in the spotlight as well.


Landon's name in big letters, "Lots of Love for Landon," always left me with a bit of a feeling of discomfort, especially after Jordan was born. Jessica Britt Jondle reminded our community from time to time that people are not an inspiration because they have a disease; that always stuck with me. Landon is not cystinosis, that is just a part of Landon, and I never wanted that to be his identity and never wanted his brother to feel as if he were not as special because he does not have cystinosis.

For the last few years, Landon has expressed interest in being in some kind of spotlight, evolving more over the last few years. It probably has something to do with creators on social media that make a lot of money and having a public image, but it's something that has appealed to him. He came home from school a few months ago and immediately logged onto his school website and asked me to sign him up for his middle school musical try-outs. I did so with a bit of hesitation, as he has never sang or danced in front of anyone and has absolutely no training, but that didn't matter to him. He passionately rehearsed his two songs and monologue with support from not only his dad and I, but his brother. Jordan is really the best cheerleader that you could meet and brags about his brother fiercely! He earned a small role in the musical and we witnessed him demonstrate such passion and excitement with his newfound interest.

My heart is so full watching him, not only because I love musicals, but because he is stepping into a spotlight, on purpose, and is shining. It made me feel curious about why this feels so joyful to me and why I don't have the same ambivalence. What came to me was a line that I recall from "Harry Potter" in which Harry says that he doesn't want people to die for him, and Ron states that people aren't dying for him, but for a cause that's much bigger. That's it. The fundraisers we have done for the CRF are not about Landon; Landon is in the spotlight because he is a representation of cystinosis in our community. It's not about Landon, or our family, but is so much bigger.

Landon, and the rest of the cast at Chartiers Valley Middle School, shined in their spotlight last weekend. Unfortunately, Landon became sick during the week of the musical, which impacted how many shows that he was able to participate in and how he felt during the shows in which he did perform. As always, Landon continues to thrive despite challenges. He performed when he could, set the necessary limits to take care of his body, and touched those around him because of his hard work, sense of humor, and positive attitude.

# COMING IN HOT

By Karolis Schroëder
HEBY, SWEDEN

MY name is Karolis and I will soon be 15 years old. I live in Heby outside of Uppsala in Sweden with my mum Annica, my little brother, Linus, and our two cats, Kajsa and Trix.

When I was 4.5 years old I was diagnosed with cystinosis and since then I have taken loads and loads of medicine. Most days it is quite okay, but I hate the fact that I always have to get up early every morning for the first dose of medicine which means I can never have a sleep-in. Well, I guess my mum does not like it very much either since I go back to sleep and she stays up with the cats and my little brother who wakes up by the sounds I make.

I try not to let cystinosis set any limits, but it is hard sometimes. I have chronic kidney disease and I think I will need dialysis or a transplant in a couple of years. I am very tired and my legs hurt from muscle wasting. Sometimes these things makes it hard for me to keep up with my friends and that makes me sad. The hardest thing besides all the medications is that I am so short and need to take a hormone shot in my leg every night and I need to go to the hospital for checkups so often.

I love playing video games, hanging out with friends and going to amusement parks and water parks. My favorite foods are hot noodles, chicken wings from the airfryer with really crisp skin and woks with a lot of chili and ginger. My friends do not understand how I can eat my food so spicy!

My dream for the future is to finish school and then work with animals or be an actor, preferably in Hollywood. I am scared that my health will prevent me from doing these things, so I hope for a cure!

KATHLEEN ROBERTS

## WORKING AS A NURSE HAS MADE ME A BETTER PERSON

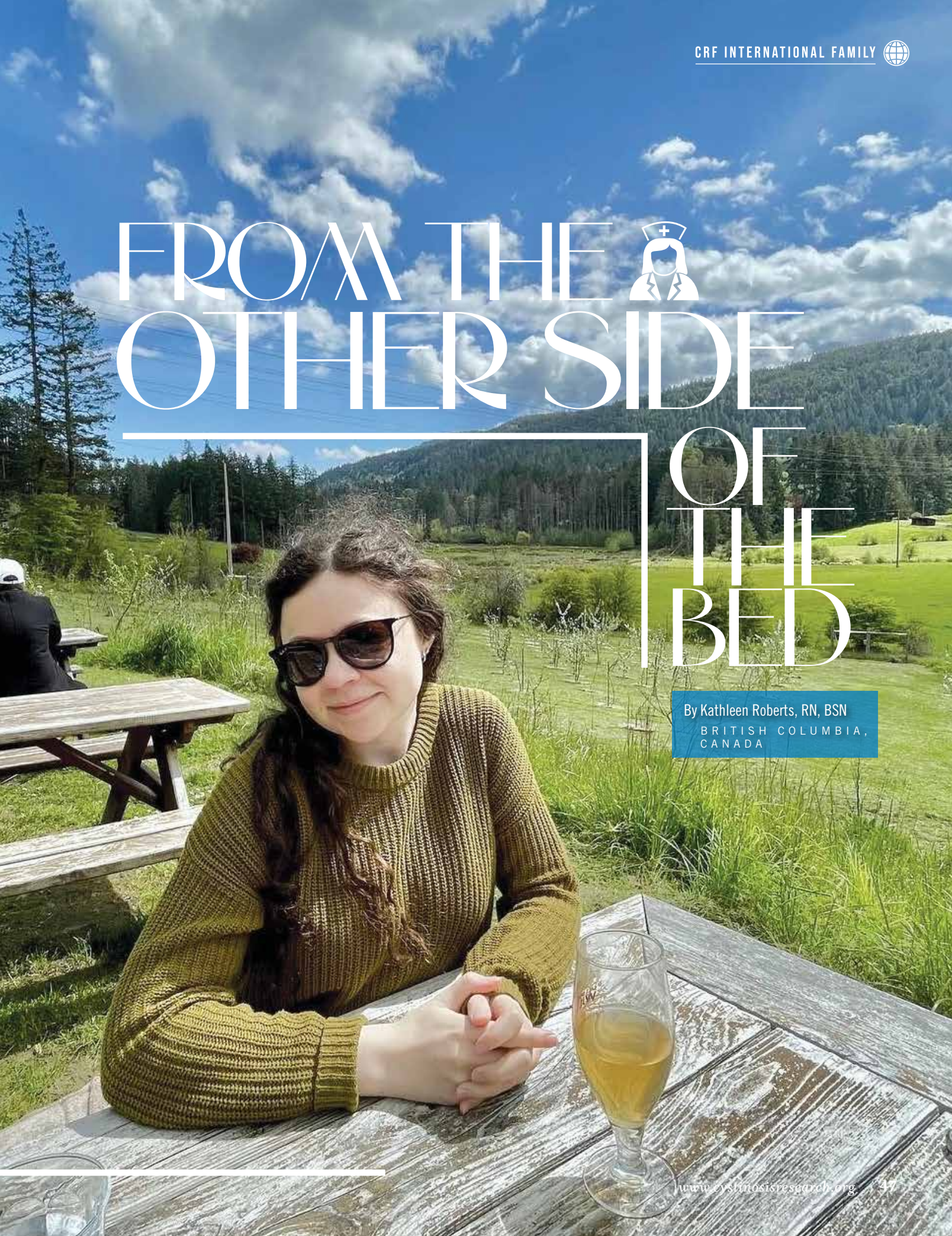
In Spring 2022, I gained five extra letters to my name: RN and BSN. I graduated from the University of British Columbia with my Bachelor of Science in nursing, passed my board exams, immediately entered a full-time position as a registered nurse in Paediatrics and soon transitioned into the Neonatal Intensive Care Unit (my absolute favourite area). As an adult living with cystinosis, this felt about as full circle as it gets. Since I was diagnosed at almost exactly one year of age, I don't remember a time that I wasn't living the patient experience. Throughout nursing school, and especially now, I have lived and am living the experience of the other side of the bed.

There is dichotomy in being both someone living with a chronic illness and a full-time working RN, caring for and responsible for many people and families like myself. There are times when I feel my contrasting experience more. It can sneak up on me. I'll be performing a skill or providing teaching to a family and suddenly I feel like the hospitalized little kid, or teen, I once was. The taste of icy hospital water through a Styrofoam cup and plastic straw immediately brings me back. A physician who was once my doctor is consulted and I see their name in the report. I flush, hot and nervous. The first time I put medications through a G-tube I had the strangest feeling as though I was on the opposite side of my childhood. Other times I am the patient and have to leave my RN role behind, let others care for me and trust their skills. At this point, I find doing so difficult.

In some ways my personal medical experience has been helpful in my nursing education and training. Pathophysiology is easier to learn when you relate disease processes to personal experiences. Pharmacology is also easier when you've taken many of the medications yourself. I knew the purpose of PICC lines, Hemovac drains, G-tubes, peripheral IVs and Foley catheters. Having a chronic illness gave me more empathy. Yet, these experiences are a drop in the bucket for the nursing profession as a whole. **Nursing is a mixture of art and science, requiring intense critical thinking abilities, an intricate understanding of biology, skill for hands on tasks, emotional intelligence to work with people in challenging situations and the ability to respond as the first point of contact for anyone involved, from family, doctors and pharmacists to techs, social workers, OTs, PTs and dieticians.** No amount of personal experience can fully prepare you.

As a nurse I have witnessed new lives come into the world and tended to someone's loved one after death. I have cared for a grieving family whose eldest child had passed away the previous night from the same infection their youngest was just then hospitalized with. I have supported mothers as their babies were apprehended and soothed numerous newborns going through withdrawal after being exposed to substances in utero. I have held and cared for tiny 2 lb. neonates and made toddlers on respiratory support both laugh and scream in fear as soon as they see me approach in the yellow isolation gown. I have had to support suicidal youth and help insert NG's into resistant eating disorder patients. At times these experiences create intense personal emotion which can feel compounded by my own experiences.

My personal healthcare journey has made me a better nurse. My ability to inform, empathize with, advocate for, relate to and support my patients is influenced by my own experience. I understand how difficult the patient side can be. But I have also been influenced by my professional experiences. From having the privilege of caring for the tiniest preemie in the NICU, the 17-year-old I had on peds or the geriatric patients I supported during my training, being part of my patients lives while working as a nurse has made me a better person, for which I will always be grateful.



# FROM THE OTHER SIDE OF THE BED

By Kathleen Roberts, RN, BSN
BRITISH COLUMBIA, CANADA

# Q + A with Hadley

By Ben Alexander, Hadley's dad
BOISE, IDAHO

For this article, I chose to interview Hadley and get her perspective on life as a 12-year-old living with cystinosis. As her dad, my viewpoint of cystinosis is through the lens of a concerned and protective parent. Over the past year, Hadley has taken on more responsibility in managing her disease, so I was curious to see how much she understands and hear her point of view. I'm proud of my daughter and the young woman she is growing into. She is kind, caring and the most empathetic person I have ever met. I hope you enjoy the following tidbits from Hadley!

**Q:** Knowing that you didn't eat solid foods until you were three, it is wild to see how far you have come in regard to the culinary world. What are some of your favorite items to eat?

**A:** Ramen, Sushi, Takis, Bahn Mi and most anything spicy.

**Q:** How often and when do you take your meds?

**A:** Three times a day, 7:30 a.m., 3:30 p.m. and 11:30 p.m.

**Q:** How do you know when it is time to take your meds?

**A:** I have alarms set on my phone to remind me when to take my pills. I am a heavy sleeper, so my dad is in charge of giving me my 11:30 p.m. meds so I won't sleep through taking them.

**Q:** How do your medications make you feel?

**A:** My pills often make me feel sick and nauseous, but it has gotten better over the years. I don't usually have a problem when I take them.

**Q:** What are your thoughts on your upcoming transition from elementary school to junior high?

**A:** I am really excited to go somewhere new and to meet new people. I have been at my current school since kindergarten and I will be a bit sad since very few of my friends will be going to my new school.

**Q:** How do you like to spend your free time?

**A:** I like to spend time with family and friends, watching movies or just talking.

**Q:** What do you want to do when you grow up?

**A:** Any job that would allow me to help people or animals and travel.

**Q:** Where do you want to go to college?

**A:** Either Boise State University or University of Idaho; I want to stay close to home.

**Q:** How do you picture your adult life?

**A:** Well, that is hard since it is so far away, but I want to stay close with my family and friends.

**Q:** What do you deal with as a result of cystinosis that you don't like?

**A:** Not being able to keep up with others. I tire easily and running is hard for me. I also don't like getting sick.

**Q:** Is there anything about cystinosis that you are grateful for?

**A:** I love to travel to the Day of Hope conference every year and to meet new people that understand what I am going through.

**Q:** Who or what inspires you?

**A:** My older sister, Stella. She is brave and strong and I can go to her with anything I need advice on, even though she can often be a brat.

**Q:** What type of music are you into and what artists do you currently like?

**A:** Taylor Swift, The Beatles, and most any '80s music.

**Q:** What is the last book you read, and did you enjoy it?

**A:** "The Tale of Despereaux." I did like it; it was different from most of the books I read but had multiple stories that tie together toward the end.

**Q:** What is one of your guilty pleasures?

**A:** Dating TV shows like "Love is Blind."

**Q:** What is one thing that can instantly make your day better?

**A:** My little cousins, Otis and Beni.

**Q:** Who is your celebrity crush?

**A:** That is hard, there are so many. I guess I would say Lewis Partridge from "Enola Holmes" and Jonathan Daviss from "Outer Banks."

**Q:** What is your biggest pet peeve?

**A:** When people chew with their mouth open.

**Q:** What is your most prized possession and why?

**A:** My dog Sherman because I love him.

**Q:** What would you do if you won the lottery?

**A:** I would donate half to charity (CRF) and the rest for college.

**Q:** What is your favorite movie genre?

**A:** Horror. I really love the movie "IT"!

**Q:** What would you tell younger kids with cystinosis to help them with the struggles of living with the disease?

**A:** Keep pushing through even though it might be hard and drink lots of water.

**Q:** Do you have any hidden talents?

**A:** Singing.

**Q:** If you could time travel to any period, what would you pick and why?

**A:** The '80s because I love that era.

**Q:** Do you have any nicknames?

**A:** Haddie, Hads, Haddie Boo, Boo, Boobalah and Josiah.

**Q:** What stresses you out?

**A:** Deadlines.

**Q:** How do you deal with stress?

**A:** I try to breathe and listen to calming music.

**Q:** What do you think about the potential cure for cystinosis and the stem cell gene therapy trial?

**A:** I'm so happy for the patients who have already been transplanted and hope I have the chance to do it in the future.

# TOGETHER, WE ARE one

**1 PURPOSE. 1 JOURNEY. 1 CURE.**

## COMMUNITY NEWS

The following pages celebrate the events dedicated to awareness and a cure by our cystinosis community. Together, we are stronger. Together, we are one!

# TOGETHER, WE ARE one

1 PURPOSE.
1 JOURNEY.
1 CURE.

## HELP CRF CONTINUE FUNDING LIFE-SAVING RESEARCH!

Did you know you can help support CRF research by planning a fundraiser? Organizing an event on behalf of someone you love with cystinosis is not only gratifying, but it also gives others a chance to support a cause that is important to you. Fundraisers can be as simple as setting up a lemonade stand or hosting a bake sale, or something more involved like organizing a golf tournament or dinner gala. When you fundraise, every dollar raised for CRF goes directly to research and every dollar counts!

A lot has changed since CRF was established. With your help, CRF has been the driving force for all cystinosis research and advances in treatment. Together, we have made an impact and have given hope to those with cystinosis for a better quality of life. When CRF started in 2003, there were only a few active research studies and no ongoing funding sources, but today CRF funds new studies twice a year and research around the world. CRF changed the course of cystinosis research and changed lives, but there is more to learn and more discoveries to be made - we need your help!

CRF is here to support you with planning a fundraiser and provide resources and tools to get you started. Every dollar you donate goes directly to support cystinosis research. Please visit the CRF website for guidance, CystinosisResearch.org/Tools-for-Fundraising/ and remember we are here to help you through the process. Thank you for your commitment and dedication to supporting our cause and joining us as we fund research for improved treatments and a cure for all those with cystinosis!

CYSTINOSISRESEARCH.ORG/TOOLS-FOR-FUNDRAISING

The Sos-Finucane Family – Erin, Matthew, and Sofie – Philadelphia, Pennsylvania

## TEAM SOFIE

Team Sofie had an amazing day on Sunday, November 20th. Erin Sos-Finucane, Sofie's mom, successfully completed the Philadelphia Marathon and for each mile she ran, she honored a person with cystinosis. Team Sofie was launched in honor of 4-year-old Sofie, to raise money for research and awareness about cystinosis. The Team Sofie community raised over $32,206 for cystinosis research! CRF is honored to partner with the Sos-Finucane family and Team Sofie. Together, we are making a difference and funding research to cure cystinosis. Thank you, Team Sofie and the Philadelphia community that supports your efforts!

TEAM Sofie

# TOGETHER, WE ARE one

1 PURPOSE.
1 JOURNEY.
1 CURE.

The Emerson Family – Jill, Clay, and Brooke – Hammonton, New Jersey

## FISHING FOR BROOKE'S CURE

The Eighth Annual Fishing for Brooke's Cure Fundraiser held in March was a huge success. This year, the fish cooperated with a record number caught and released. The experienced anglers landed a record number of 496 fish which ensured another successful fundraiser for the Emerson Family. We are grateful to your dedicated group of friends, family, and community for their support that raised nearly $35,000 in honor of Brooke and cystinosis research.

Thank you, Jill, Clay and Brooke, for hosting and organizing this extraordinary fundraiser and your commitment to CRF. Since your first Fishing for Brooke's Cure event in 2015, CRF has received $222,728 to fund cystinosis research. With your help, we are changing lives and providing a brighter future for our children and adults with cystinosis!


The Beauregard Family – Courtney, Kevin and Lily – Swansea, Massachusetts

## FOURTH ANNUAL CHILI COOK-OFF

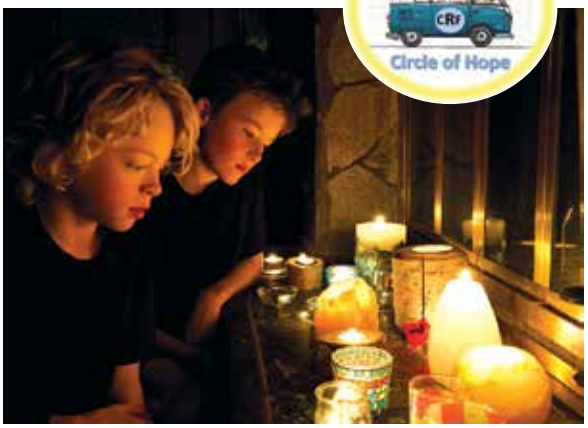
The fourth Annual Chili Cook-off for Cystinosis Research was held in December 2022, in Fall River, Massachusetts, in honor of Lily Beauregard. The successful event was organized once again by Shelli Pereira, a dear friend of the Beauregard family. An enthusiastic community attended to support Lily along with 18 creative chili cooks, a group of dedicated volunteers, entertaining musicians, and very generous donors, and best of all, Lily was healthy enough to attend this year! The successful festivities and support resulted in $5,900 raised for CRF and cystinosis research. Thank you to the Beauregard family, their friends, and neighbors for their generosity and support in providing a brighter future for Lily and all the children and adults affected by cystinosis. We are grateful to Shelli for her time and commitment and for already planning the Fifth Annual Chili Cook-off for Cystinosis Research in 2023. Thank you!

Help Lily BLOSSOM

# TOGETHER, WE ARE one

1 PURPOSE.
1 JOURNEY.
1 CURE.

The deBruyn Family
Kristen, Nathan, Seth and Leif
Calgary, Alberta, Canada



## SETH'S CIRCLE OF HOPE

Each year on October 21, Kristen and Nathan deBruyn mark the anniversary of their son Seth's cystinosis diagnosis by lighting a candle, creating a Circle of Hope. Today, Seth's Circle of Hope candle-lighting remembrance has spread throughout the cystinosis community and beyond, sending a message of hope throughout the world.

"Wherever you are in the world, please join us in our Circle of Hope as we re-kindle our strength, hope and commitment to finding a cure for cystinosis. We welcome your presence as we express our appreciation for the many gifts that life brings; for the love, friendship and hope that abounds." - Kristen Murray

The simple act of lighting a candle reminds all of us that hope abounds. In 2022, the deBruyn annual candle lighting ceremony raised $25,154 for cystinosis research. CRF's partnership with the deBruyn family and the Canadian Cystinosis Research Foundation ensures that donations will be used for CRF funded research projects.

Thank you to the deBryun family for supporting cystinosis research. Please mark your calendars for October 21, 2023, and light a candle of hope for our children and adults with cystinosis.

The Galloway Family
Christina, Hunt, Rowyn and Collins
Cumming, Georgia


## SECOND ANNUAL COLLINS' CURE CUP

Thank you to the Galloway family for hosting their 2nd Annual Collins' Cure Cup Golf Tournament at the Windermere Country Club in November 2022. The event was a tremendous success thanks to the tournament sponsors, dedicated volunteers and 114 enthusiastic golfers. Following the fun-filled day of golf, the Cumming community of family and friends joined the participants for dinner, live music and fundraising festivities. We are grateful to the Windermere Country Club and its staff and especially the Cumming community for their contributions and support of the Galloway family. Because of your generosity, CRF has received over $53,300 in honor of Collins and cystinosis research. Thank you for making a difference in the lives of those with cystinosis!

## Cure Cystinosis International Registry

# Our Link Between Patients and Researchers

**By Clay Emerson, PhD, PE, CFM**

Brooke Emerson's dad, CRF Trustee, and CCIR Committee Member

*Hammonton, New Jersey, USA*

**The new** Cure Cystinosis International Registry (CCIR) was launched in the Spring of 2021. The new registry features revised questions reflecting the ever-evolving treatment of the disease. Patients and caregivers who may have completed the original registry (pre-2021) are encouraged to register for the new registry. Participants are also encouraged to update their responses periodically as their treatment and symptoms may change over time.

In just the first two years, patients from 13 countries have participated in the registry. Participants in the registry also include patients representing 22 states across the United States. The registry provides a critical link between patients and researchers. Due to the ultra-rare nature of the disease as well as the myriad of complications the disease presents, progress towards improved treatment and an ultimate cure for cystinosis can only be possible with the valuable input from our limited patient community.

## Quality of Life

As a multisystemic disease, cystinosis impacts numerous organs throughout the body, with the first symptoms usually resulting from early damage to the kidneys. The CCIR has confirmed that most cystinosis patients take 10 or more medications with an around-the-clock dosing schedule. Compounding these issues is the fact that many current medications require rigorous dosing schedules and come with a multitude of side effects and undesirable drug interactions. Between the multitude of impacts and the plethora of medications, what can often be overlooked is the serious quality-of-life impacts that come along with the disease and its management.

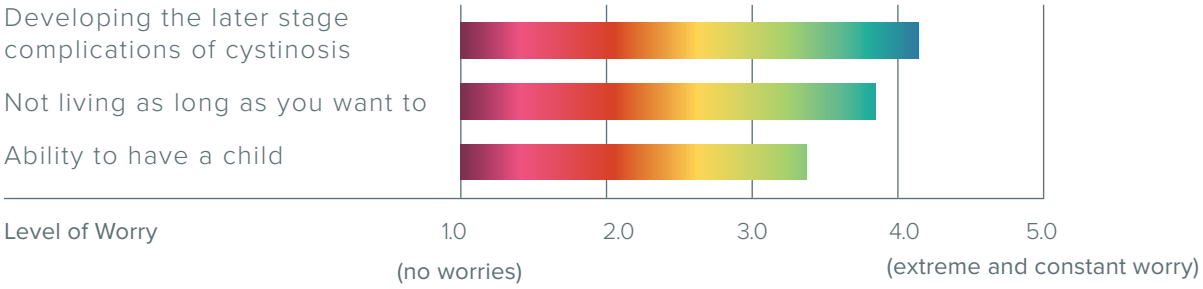
***EARLY DATA FROM THE CCIR ILLUSTRATES THE PREVALENCE OF QUALITY-OF-LIFE IMPACTS ASSOCIATED WITH CYSTINOSIS***

Early data from the CCIR illustrates the prevalence of quality-of-life impacts associated with cystinosis for both the patients and their caregivers. Cystinosis and its treatment often impacts and interrupts a child's education and presents similar challenges for working adults with cystinosis. When participants were asked how often cystinosis has impacted the participant's school/work attendance, nearly 60% responded either Sometimes, Often or Always.

People living with chronic disease conditions like cystinosis often worry about many things. In the new CCIR patient registry, participants and caregivers were asked to score how much the participant or the caregiver worries about various quality-of-life issues, rating each item from 1 (no worries) to 5 (extreme and constant worry). The three items representing the highest average respondent scores illustrate some of the primary day-to-day challenges and concerns of participants.

Cure Cystinosis
International Registry

HOW MUCH DO YOU WORRY ABOUT VARIOUS QUALITY-OF-LIFE ISSUES?




**In the** first two years since its launch, the new cystinosis patient registry is already helping to inform researchers and ultimately accelerate the development of better treatments and a cure for cystinosis. The value of the registry will only truly be realized with patient participation. We strongly encourage patients or caregivers to participate in the registry and help identify the needs of patients with cystinosis and ultimately accelerate research. The questionnaire takes about 40 minutes to complete, and registration is simple. Please visit the CRF website to sign up today!

***WE STRONGLY ENCOURAGE PATIENTS OR CAREGIVERS TO PARTICIPATE IN THE REGISTRY***

**www.cystinosisresearch.org/cure-cystinosis-international-registry**

# Stéphanie Cherqui, PhD

**PROFESSOR, DEPARTMENT OF PEDIATRICS, UNIVERSITY OF CALIFORNIA SAN DIEGO**

## NAMED TOP 23
## WOMEN IN ACADEMIC ENTREPRENEURSHIP


**Behind every transformative technology is a team of brilliant scientists and researchers.** Academic research in particular is a driving force for technological innovation. Here, we have identified a "Midas List" of researchers who have pioneered highly transformative technologies that have translated to the clinic. Using venture-backed startups as a primary guide along with other impact metrics such as patents, citations and thought leadership, the following professors have demonstrated patient impact, providing significant contributions to the startup ecosystem.

The Life Sciences ecosystem is experiencing unprecedented innovation, and academic entrepreneurs are at the forefront of this change. Women's History Month is a time to celebrate women from past and present and all walks of life. In honor of International Women's Day 2023, we at BIOS have the pleasure of putting together the "Top 23 Women in Academic Entrepreneurship" that have truly changed the life sciences ecosystem as we know it.

Stéphanie Cherqui is a Professor of Pediatrics and Chair of the UC San Diego Cystinosis Stem Cell Gene Therapy Consortium. A pioneering expert in cystinosis, Cherqui and her research group, the Cherqui Lab, focus on the use of stem cell and gene therapy for multisystemic genetic disorders and fundamental understanding of tissue repair by bone marrow stem cells. Cherqui is currently developing the first stem cell gene therapy clinical trial for cystinosis at UC San Diego. Cherqui is the co-founder of Stelios (Acquired by Lexeo Therapeutics) & Papillion Therapeutics.

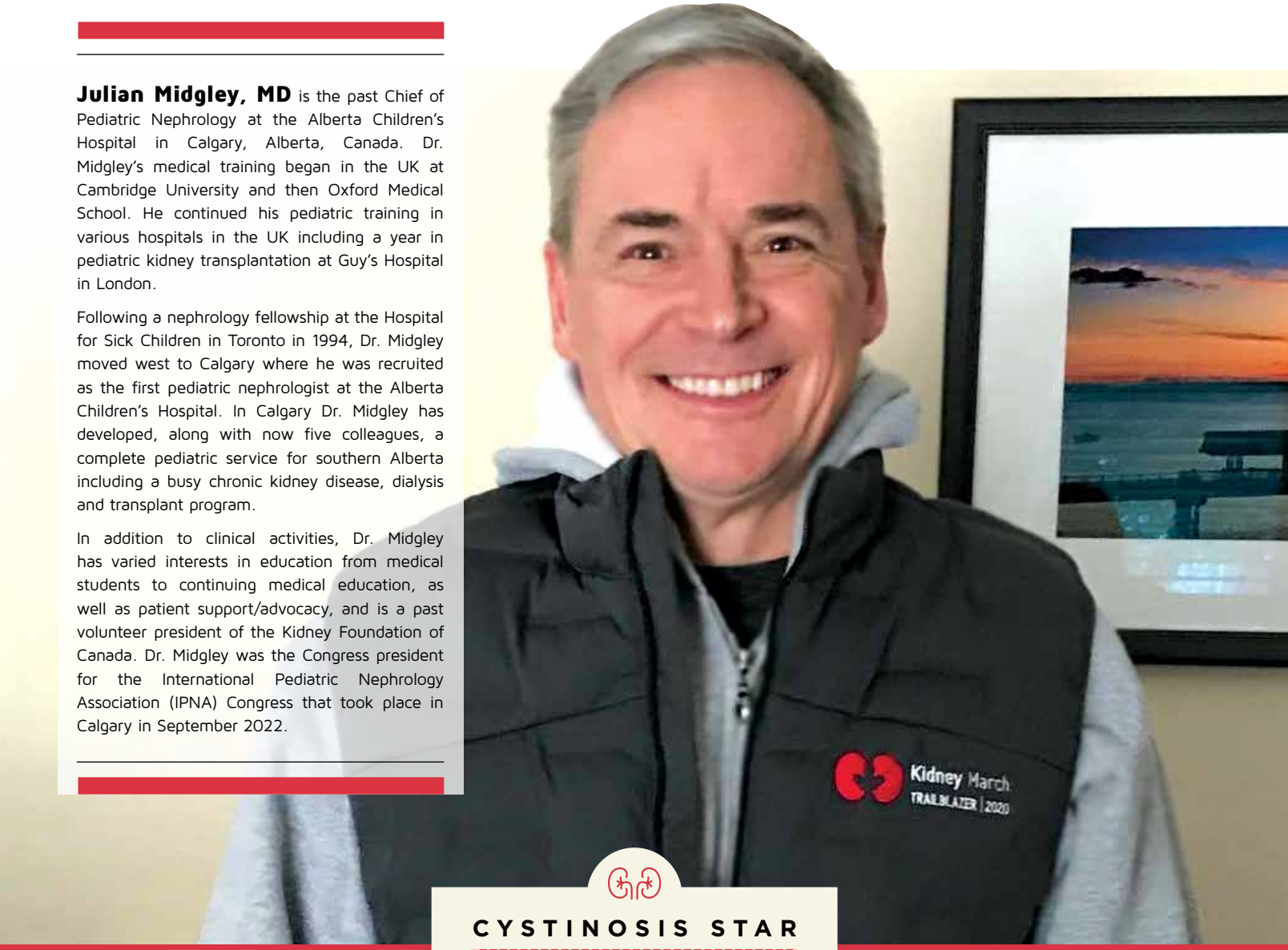
20 years TOGETHER

# RESEARCH UPDATES

**Julian Midgley, MD** is the past Chief of Pediatric Nephrology at the Alberta Children's Hospital in Calgary, Alberta, Canada. Dr. Midgley's medical training began in the UK at Cambridge University and then Oxford Medical School. He continued his pediatric training in various hospitals in the UK including a year in pediatric kidney transplantation at Guy's Hospital in London.

Following a nephrology fellowship at the Hospital for Sick Children in Toronto in 1994, Dr. Midgley moved west to Calgary where he was recruited as the first pediatric nephrologist at the Alberta Children's Hospital. In Calgary Dr. Midgley has developed, along with now five colleagues, a complete pediatric service for southern Alberta including a busy chronic kidney disease, dialysis and transplant program.

In addition to clinical activities, Dr. Midgley has varied interests in education from medical students to continuing medical education, as well as patient support/advocacy, and is a past volunteer president of the Kidney Foundation of Canada. Dr. Midgley was the Congress president for the International Pediatric Nephrology Association (IPNA) Congress that took place in Calgary in September 2022.




CYSTINOSIS STAR

# THE AMAZING, REMARKABLE, MARVELLOUS... DR. JULIAN MIDGLEY

**by Kristen Murray,** CRF Board Member and Seth deBruyn's mom

**THE** recipient of the Canadian Society of Nephrology Distinguished Service Award in 2018, Dr. Midgley's medical practice is indeed distinguished. As if providing impeccable, holistic patient care weren't enough, Dr. Midgley is abundant in his offering of kindness, empathy and support to patients and their families alike. A staunch advocate for his cystinosis patients, Dr. Midgley plays an active role in the broader cystinosis community and with the Cystinosis Research Foundation. He sits on the CRF Medical and Scientific Advisory Board and is a regular and much anticipated presenter at annual CRF Day of Hope family conferences. Dr. Midgley has treated 23 patients with cystinosis to date, and patients and their families are quick to describe him as brilliant, respectful, meticulous, methodical, proactive, nuanced, kind, playful and, of course, very funny.

Dr. Midgley offers an abundance of kindness, empathy and support for patients and their families alike.

As a parent to Seth, who has cystinosis and is under Dr. Midgley's astute care, I know first hand the exceptionally high calibre of Dr. Midgley's care. Yes, Dr. Midgley is the best, our favourite, a hero, really, who makes our everyday life with cystinosis more manageable and so much brighter than it would otherwise be. Offering the most collaborative, thoughtful care that we as a family have ever experienced, Dr. Midgley creates a space in his clinical practice that we happily anticipate and truly enjoy. At appointments, Dr. Midgley enlists our ideas and opinions, asks our preferences and includes us in decision-making. He responds — yes, responds — night and day to our email queries about medication, lab results and a myriad of pressing questions. He provides emergency, out-of-province prescriptions for forgotten medications on holiday sojourns (ahem...first hand experience). He happily poses in photos, willingly supports and even speaks at family fundraising events and jauntily hops on board for interviews like this...

Dr. Midgley with young Seth deBruyn.

Jordan Janz, the first Stem Cell and Gene Therapy clinical trial participant, with Dr. Midgley.

*As I was preparing for my talk with Dr. Midgley, 11-year-old Seth and his twin brother Leif asked me to include a few questions of their own to ask Dr. Midgley...important questions of course. And so I started off with these, and Dr. Midgley was happy to oblige.*

### WHAT IS YOUR FAVORITE COLOUR?

I wear a lot of blue, but I actually prefer red. I have a red stethoscope and my jacket is red.

*I had noticed Dr. Midgley was indeed wearing a red windbreaker jacket. This jacket was complemented by a well-worn pair of hiking boots and a robust-looking waterproof biking pannier. And so, I had to ask...*

### WE KNOW THAT YOU BIKE TO WORK. BUT EVEN IN WINTER...CANADIAN WINTER?

Yes, I've always ridden to work. I rode to grammar school and university, toured Italy on a tandem bike, in fact, and rode to work in Toronto. I don't bike if there is too much snow on the ground, and not if it is colder than -10 degrees Celsius or so.

### DO YOU HAVE STUDDED TIRES FOR THE ICE?

Oh no, I just get off and walk on the slippery sections and it seems to work quite nicely.

### HAVING NOTED YOUR BRITISH ACCENT AND ORIGINS, THE KIDS WANT TO KNOW... DO YOU PREFER SOCCER OR CRICKET?

When I was younger our parents used to take us to cricket test matches. When I was at the equivalent of high school in the UK, the school played cricket in the summer and rugby in the winter. I loved cricket. I wasn't very good, but I was able to catch the ball very well, so I did some very good catches. I continued to play cricket when I was in university at Cambridge and it was good fun.

>>>>

>>>> CONTINUED

### WHAT IS YOUR FAVORITE FOOTBALL (SOCCER) TEAM?

The Hammers is my favorite team. Yes, that's the nickname of West Ham United in the east of London. My twin brother supported Spurs, the nickname for the Tottenham Hotspur Football Club, which is in the north of London. So there was some good rivalry.

*(And as the mom of twins, I had to ask:)*

### WHAT WAS IT LIKE GROWING UP AS A TWIN?

It's hard to say what it was like being a twin, because I didn't know anything different. It was good to have a constant companion with whom to play and bike and go to school. We went to the same schools all through high school, but then he went to Oxford and I went to Cambridge, and he is now an engineer.

### CATS OR DOGS?

When I was growing up, we had a beagle named Emma, and beagles are a menace because they eat anything — socks, shoes, etc. One of my favorite memories is walking Emma with my dad. It was good time spent with my dad.

### YOU CARE FOR SO MANY CHILDREN. TELL US A LITTLE ABOUT YOUR OWN KIDS.

I have 4 kids ages 27, 29, 32 and 37, Ben, David, Ruth and Claire. Ruth just had a baby girl in February, so I am a new grandfather, which of course is very exciting.

### WHAT ARE SOME THINGS THAT WE MIGHT NOT KNOW ABOUT YOU?

I play squash. I started playing as a teenager and was the captain of the squash team at my college in Cambridge. I am also the team manager of an under-21 junior hockey team.

*And so, having covered all of my kids' questions, we moved to some more cystinosis-related topics:*

### WHEN DID YOUR JOURNEY WITH CYSTINOSIS BEGIN?

I encountered a few patients with cystinosis in the UK and during my fellowship at the Hospital for Sick Children in Toronto, but didn't have extensive interactions with them. I came to Calgary in 1994 as the first pediatric nephrologist. And so, I had a few patients who were transferred from other clinics, and one of them had cystinosis. And really, it was her disease, her experience, her story that taught me a lot about the disease. I am very pleased that I still know and treat that patient 29 years later.

### WHAT WAS IT LIKE TO TREAT SUCH A RARE DISEASE AS CYSTINOSIS?

Karen McCullagh, Kristen Murray, Dr. Midgley and Deb Pym.

Well, beyond using my "book knowledge," I reached out to my previous colleagues and teachers, and I connected with them a lot. Dr. Paul Goodyear, in Montreal, became a contact. Bill Gahl, medical geneticist with the National Institute of Health in the US also became an important contact. He visited Calgary within the first few years of my time there and was an important connection indeed.

### HOW DOES TREATING CYSTINOSIS COMPARE TO THE TREATMENT OF OTHER DISEASES?

There are similarities in treating any chronic illness that come with the longevity of care, and the opportunity that this provides for developing relationships with patients and their families. Chronic kidney disease is systemic. It affects all sorts of things in the body and has so many implications for bodily functioning. And cystinosis is like that, but even more complex and a magnitude more severe because it is so relatively significant and difficult soon after diagnosis.

### HOW WOULD YOU DESCRIBE THE DIAGNOSIS STAGE OF THE CYSTINOSIS JOURNEY?

The beginning stages of diagnosing cystinosis can be a very emotional time for families. Diagnosing cystinosis is almost always difficult, with much time and struggle before, with such lack of clarity and so much confusion and stress. One family put it very well to me and they said, "We were so elated to know what the problem is, but at the same time we are so devastated to know what the problem is." The diagnosis is so difficult because cystinosis is so difficult to treat, and because up until recently, there has been no cure for the underlying disease.

### WHAT DO YOU SEE AS BEING THE BIGGEST CHALLENGES FOR PATIENTS AND FAMILIES LIVING WITH CYSTINOSIS?

I think that it depends on where you are at in the journey. The first challenge for families is often understanding and accepting what the disease is all about. Another challenge is the relentlessness of the treatment. Parents have to be so dedicated (almost obsessive) about medications. And this can be a boon of course in terms of disease treatment, but timing the medication at exact increments around the clock, day in and day out, can be such a burden on families.

### WHAT STRENGTHS HAVE YOU SEEN IN PATIENTS AND FAMILIES LIVING WITH CYSTINOSIS?

Patients with whom I work often don't see cystinosis as being a burden in their lives. They see it as having helped them to be better people, stronger people. Paradoxically, they see cystinosis as having helped them to reach their goals and to find success.

### YOU ARE WIDELY KNOWN AS A DOCTOR WHO IS EXCEPTIONALLY EMPATHIC, KIND AND SO VERY UNDERSTANDING. WHAT DO YOU THINK INSPIRES THIS "WAY" THAT YOU HAVE?

I can't imagine what it would be like to be a parent of a child who has a rare and difficult condition. I really try to remember that it is the family's first time in their journey with the disease. I try to see what they need at any given stage to enable them to do what needs to be done. I want to give patients and families the specific information (I love talking about kidneys) and the support they need to make the very best of their situation.

Supporting families to take care of their children is really what makes children's lives so much better. I see it as crucial to develop strong partnerships with parents, because parents are going to be the people doing all the "real" work. I have the delight and good fortune to develop relationships with families.


Dr. Midgley with Andrew Cunningham and family.

I look forward to seeing my patients and families and to working with them to optimize health and to get good results. It is very rewarding and I think that this makes things quite positive.

### AS A PHYSICIAN WHO WORKS WITH CHRONIC AND SERIOUS ILLNESS, HOW DO YOU DEAL WITH THE GRAVITY AND DIFFICULTY THAT YOU MEET IN YOUR PRACTICE EACH DAY?

We couldn't possibly offer the care from our nephrology clinic without the support of the nurses, pharmacists, dieticians, social workers, psychologists and colleagues. We chat about challenging issues, we collaborate, we support each other. We have strong relationships. It feels like family and this makes a real difference.

### WHEN DID THE CYSTINOSIS RESEARCH FOUNDATION FIRST COME TO YOUR AWARENESS?

Years ago, a family in my clinic told me about a conference that they had gone to in California called the Day of Hope and said that I must go. And that's the biggest regret that I have in my cystinosis journey, that I didn't go to the conference for a number of years. With only two nephrologists on staff, things were so busy that I just couldn't get away. But now, the thought of missing the Day of Hope, well, it is just such a valuable place to be, and CRF is such an important organization with which to have connection.

### WHAT IMPACTS HAVE YOU NOTICED THE CRF HAS ON PATIENT AND FAMILY LIFE?

Meeting other people, other patients in similar age groups, family members, hearing the active research about improvements to care and finding the support in the community all have such a positive impacts on patients and their families. CRF does a superb job in balancing the need for research and the need for patient support. Research is of course, very important. But it is only important to families if it enables them to have hope for an improvement in their child's life. The way that CRF grounds its research program in the patient perspective and needs really leads the way.

### WHAT IS IT LIKE TO BE PART OF A RARE DISEASE COMMUNITY FOR WHICH THERE IS A CURE ON THE HORIZON?

When I first heard about stem cell treatment for cystinosis, I was sceptical at first. But then came the realization that the cells that have the modified gene can transfer cystinosin to other cells in the body, and this was remarkable. In seeing images of nano-tubules and animal models showing benefits from a stem cell transplant, I became more hopeful about the possible benefit beyond the bone marrow and into the whole body.

The idea of a cure has a real enormity of possible benefit to patients and families. It is such a substantial move forward. Stem cell transplant is a challenge, of course; it is a big deal. But the idea of having one treatment, albeit difficult, to make a substantial change in cystinosis is lightyears ahead of taking medication two, three, four times or more a day.

### WHAT ARE YOUR THOUGHTS ABOUT MEDICATION COSTS?

One of the things that really bugs me about treating patients with cystinosis is the financial issues that families face. It really bothers me that medications for rare diseases tend to be so very expensive. Fortunately in Canada, the provincial health care systems usually cover the costs of expensive medications such as cysteamine with government-sponsored medication insurance. We are so fortunate in this respect. However other treatments, for example electrolyte supplements, that are absolutely crucial to treat cystinosis, are not always covered by private or government sponsored insurance plans and can be quite costly at the doses needed by patients with cystinosis. I would love to have the costs of all required medications covered so that families don't have to take on that extra burden.

### WHAT ARE SOME UNIQUE ASPECTS OF CYSTINOSIS CARE THAT YOUR TEAM AT THE ALBERTA CHILDREN'S HOSPITAL OFFERS?

We are so pleased to offer continuity of patient care into adulthood, an aspect of care that is very helpful and important. It allows us to maintain the strong relationships that we have with patients and families. It can be so difficult for patients to change their health care provision when they turn 18, as so many other aspects of their lives are changing. I am also so very pleased to be involved in the care for patients with cystinosis who choose to have children.

### WHAT ARE YOUR HOPES FOR THE FUTURE OF CYSTINOSIS TREATMENT?

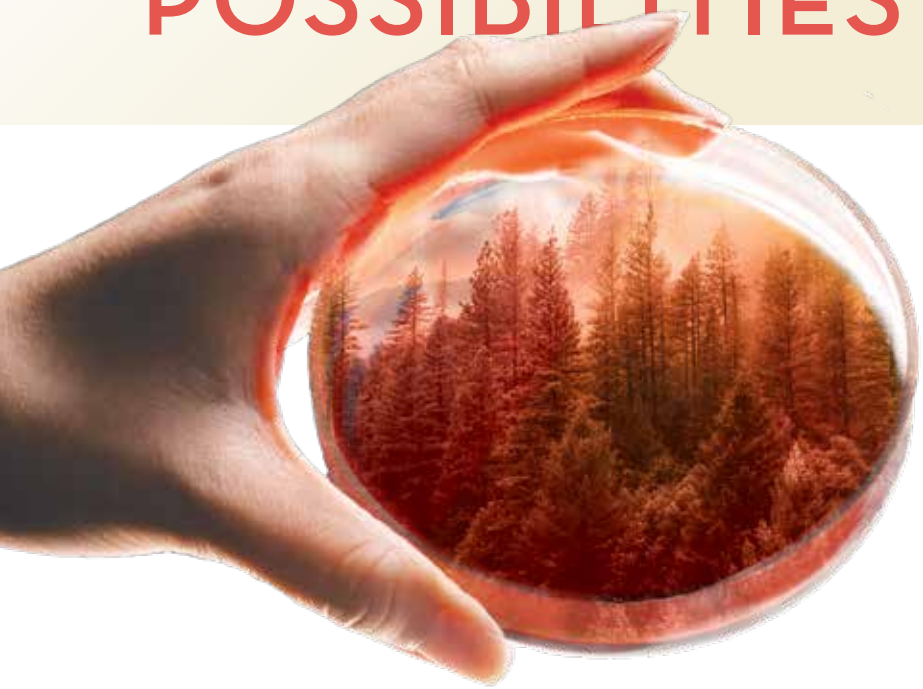
I would like to have the stem cell transplant available in Canada, ideally with Calgary as the site. I would also like to see the initiation of a neonatal screening program. Yes, I would certainly like to see these ideas come to fruition.

*Dr. Midgley, thank you so much for talking with me. Thank you for taking such impeccably good care of your patients and their families, for being such a vibrant member of the cystinosis community and for shining such tremendous light on our paths. We are grateful for you.*

Thank you, too. It is my pleasure.

Jordan Janz and his mom Barbara Kulyk with Dr. Midgley and Jenny Wichart.

# IN A SMALL DISH, POSSIBILITIES MULTIPLY



***By reprogramming cells donated by cystinosis patients, Dr. Benjamin Freedman is creating new pathways to kidney health, including progress toward the breakthrough goal of regeneration.***

by Dennis Arp

**IT** all starts with a humble urine sample. From such simple beginnings, breakthroughs are born in the cutting-edge research lab of Dr. Benjamin "Beno" Freedman, whose work on kidney regeneration holds the promise of translational advances for cystinosis patients.

By reprogramming cells captured from those urinary samples, Dr. Freedman and his team create stem cells and kidney grafts to see how they respond to various kinds of cystinosis therapy. They want to reproduce classic symptoms of cystinosis so they can better understand the causes of the disease and why kidneys are so vulnerable.

Like the cells and grafts themselves, the possibilities are growing.

"The other side of this work is to push forward on regeneration, so that even if the kidneys of cystinosis patients get sick, we can discover tools for making healthy kidneys," says Dr. Freedman, Associate Professor of Medicine in the Division of Nephrology at the University of Washington.

Kidney regeneration from tiny cells in a petri dish may sound like far-off science fiction. But thanks to funding support from the Cystinosis Research Foundation, Freedman's work is already achieving tangible results, with the very real potential of more insights to come.

"We're in the fourth year of funding from CRF, and it's been instrumental in our progress," Dr. Freedman says. "The support has gotten us off the ground in a big way. It keeps us focused, and it allows us to do experiments that are bolder than traditional grant mechanisms would allow."

## A LONGTIME COMMITMENT TO INNOVATION

Dr. Freedman has never been afraid to go bold in his stem cell research and his pursuit of innovation in regenerative medicine. He first started down this research path more than 15 years ago, with his focus turning to kidney-related ailments in 2008 when the health of a beloved uncle was failing, forcing him to begin the journey to a second kidney transplant.

"At the time, there was no way to turn stem cells into kidney organoids, but I said, 'Hey, this could be very useful for people like my uncle,'" Dr. Freedman said.

*Next steps for the team include* "CLINICAL TRIALS IN A DISH"

Kidney organoids resemble miniature kidneys. They contain filtering cells connected to tubes, and can respond to infection and therapeutics in ways that parallel the responses of kidneys in people.

Dr. Freedman and his colleagues are motivated to gain understanding from their advances, but in the early days of their research they didn't know what would happen if they put the organoids they had created from cells into the body, and they didn't know at what stage of the process that transplantation would be effective.

"One of the real highlights of these research projects CRF is making possible is that we've been able to see what these cells can do and the extent to which they can form new kidney tissue," Dr. Freedman said.

CYSTINOSIS STAR

# BENJAMIN "BENO" FREEDMAN, PhD

***Associate Professor of Medicine***

University of Washington, School of Medicine

## DISEASE-IN-A-DISH MODEL YIELDS INSIGHTS

"Along the way, there have been many challenges, not the least of which is getting disease symptoms of cystinosis to manifest in a petri dish," Dr. Freedman said.

"We made the cells, but it wasn't immediately obvious that those cells had cystinosis," he noted. "Even though they had the right genotype, they didn't show the phenotype – the recognizable symptoms. It took some extra work for us to figure out how to coax out the disease. Getting a disease-in-a-dish model has been very gratifying. It has taught us quite a bit about the general disease process."

Now that the tools of their research project are built, Dr. Freedman and his colleagues have reached a stage where "we can put them all together, we can use them in many different ways and we can share them with other researchers," he said. "This is a very exciting time."

The cell lines they're creating essentially constitute a new biobank of samples that can be used in the study of cystinosis for decades to come.

"We're especially interested in utilizing the cells to understand how different patients respond to different types of interventions," Dr. Freedman added.

## STREAMLINING THE ROUTE TO CLINICAL TRIALS

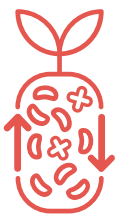
Next steps for the team include "clinical trials in a dish" – a relatively new type of investigation facilitated largely by technological advances such as induced pluripotent stem cells (iPSCs), whole-genome sequencing and CRISPR gene editing. On this research route, investigators often can move forward quickly, gaining early insights about possible treatments without the risk of involving human trial participants.

"It's something that the U.S. government has started moving to acknowledge and support – the idea that we need to move faster with trials," Dr. Freedman said.

On the horizon, they hope, is study of the cells in cystinotic mice.

"We're working on developing the mice we need, and I'm cautiously optimistic that we'll start doing transplantations within the next six months or so – as soon as the strains are well established," Dr. Freedman said.

Ultimately, if progress continues, the project could move into human clinical trials within five to 10 years. That might seem like a long way off, but thanks to the grant support of the CRF and the inspiration of cystinosis families, he and his team are in this effort for the long haul.



**"It definitely keeps me motivated to meet the families at CRF's Day of Hope Conference and also during the donor process," he said. "When I listen to their stories, it just makes me realize the level of dedication they have and the degree to which they are rising to the challenge."**

That tenacity and resilience permeate his research team. Dr. Freedman is only half kidding when he says, "I see people in our lab and I say, 'Hey, have you cured cystinosis yet? Because kids are waiting, so we've got to get a move-on.'"

# STÉPHANIE CHERQUI, PhD AWARDED $4.8 million CIRM Grant for Friedreich's Ataxia

We have always known that studying a complicated disease like cystinosis would lead to discoveries in other more prevalent diseases and disorders. On November 29, Dr. Cherqui, a professor in the Department of Pediatrics at the University of California San Diego, (UCSD), was awarded a $4.8 million grant from the California Institute of Regenerative Medicine (CIRM) for her work on Friedreich's Ataxia, a degenerative neuromuscular disorder.

It was CRF's funding of Dr. Cherqui's cystinosis work that provided the foundation for her research on Friedreich's Ataxia. To date, CRF has awarded over $6.1 million to Dr. Cherqui for her cystinosis stem cell and gene therapy work. Dr. Cherqui is appreciative of CRF's early and continuous support of her stem cell and gene therapy project which resulted in an FDA-approved clinical trial for cystinosis and a CIRM grant for a potential treatment for Friedreich's Ataxia.

"The gene therapy approach we are developing for Friedreich's Ataxia is based on the discoveries we did on cystinosis. The fact that hematopoietic stem cells could rescue a disease such as cystinosis and the mechanism by which these cells could provide the healthy protein to tissues opened new perspectives in the application of such a therapy to other genetic disorders."

Congratulations to Dr. Cherqui for this accomplishment. We are grateful for her dedication to the cystinosis community and for helping other rare disease communities.

VIEW THE ARTICLE ON THE CRF WEBSITE:

**www.cystinosisresearch.org/the-expansive-impact-of-crf-research**

EIGHTH INTERNATIONAL

# CYSTINOSIS RESEARCH SYMPOSIUM

Sponsored by the Cystinosis Research Foundation

SAVE THE DATE

## THURSDAY, SEPTEMBER 7, 2023
AND
## FRIDAY, SEPTEMBER 8, 2023

*Arnold and Mabel Beckman Center of the National Academies of Sciences and Engineering*

IRVINE, CALIFORNIA

## 2023 SYMPOSIUM CO-CHAIRS

Corinne Antignac, MD, PhD

Stéphanie Cherqui, PhD

Julie Ingelfinger, MD

BY INVITATION ONLY

cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

*For information contact CRF President Nancy Stack*
*(949) 223-7610 or nstack@cystinosisresearch.org*

# THE IMPACT of CRF RESEARCH

20 years

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

## New Drug Discovery Cysteamine, New Medications and Devices

*31 GRANTS*

**Ghanashyam Acharya, PhD**
BAYLOR COLLEGE OF MEDICINE, HOUSTON, TEXAS

**Francesco Bellomo, PhD**
**Francesco Emma, MD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Pierre Courtoy, MD, PhD**
**Christophe Pierreux, PhD**
DE DUVE INSTITUTE, LOUVAIN UNIVERSITY MEDICAL SCHOOL, BRUSSELS, BELGIUM

**Laura Rita Rega, PhD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Antonella De Matteis, MD**
TELETHON INSTITUTE OF GENETICS AND MEDICINE, NAPLES, ITALY

**Ranjan Dohil, MD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

**Francesco Emma, MD**
**Laura Rita Rega, PhD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Paul Goodyer, MD**
MONTRÉAL CHILDREN'S HOSPITAL, MONTRÉAL, QUÉBEC, CANADA

**Jennifer Hollywood, PhD**
**Alan Davidson, PhD**
UNIVERSITY OF AUCKLAND, AUCKLAND, NEW ZEALAND

**Michael Sekar, PhD**
AMMA THERAPEUTICS, INC., HAYWARD, CALIFORNIA

**Laura Rita Rega, PhD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Vincent Stanton, Jr., MD**
**Patrice Rioux, MD, PhD**
THIOGENESIS THERAPEUTICS, INC., SAN DIEGO, CALIFORNIA

## Cure Cystinosis International Registry (CCIR)

*1 GRANT*

**Ranjan Dohil, MD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

## Cellular and/or Molecular Studies of the Pathogenesis of Cystinosis

63 GRANTS

**Corinne Antignac, MD, PhD**
IMAGINE INSTITUTE (INSERM U1163), PARIS, FRANCE

**Francesco Bellomo, PhD**
**Francesco Emma, MD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Sergio Catz, PhD**
**Raquel Carvalho Gontijo, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Sergio Catz, PhD**
**Danni Chen, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Sergio Catz, PhD**
**Farhana Rahman, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Sergio Catz, PhD**
**Apama Shukla, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Sergio Catz, PhD**
**Nadia Zgajnar, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Antonella De Matteis, MD**
TELETHON INSTITUTE OF GENETICS AND MEDICINE, NAPLES, ITALY

**Olivier Devuyst, MD, PhD**
**Zhiyong Chen, PhD**
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

**Olivier Devuyst, MD, PhD**
**Alessandro Luciani, PhD**
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

**Liang Feng, PhD**
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

**Liang Feng, PhD**
**Xue Guo, PhD**
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

**Bruno Gasnier, PhD**
**Yann Terras, MSc**
CNRS/UNIVERSITÉ DE PARIS, PARIS, FRANCE

**Taosheng Huang, MD, PhD**
UNIVERSITY OF CALIFORNIA, IRVINE, IRVINE, CALIFORNIA

**Elena Levtchenko, MD, PhD**
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

**Ming Li, PhD**
**Jacob Kitzman, PhD**
UNIVERSITY OF MICHIGAN, ANN ARBOR, MICHIGAN

**Alessandro Luciani, PhD**
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

**Gennaro Napolitano, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Norbert Perrimon, PhD**
HARVARD MEDICAL SCHOOL, BOSTON, MASSACHUSETTS

**Giusi Prencipe, PhD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Matias Simons, MD**
**Zvonimir Marelja, PhD**
IMAGINE INSTITUTE, PARIS, FRANCE

**Jess Thoene, MD**
TULANE UNIVERSITY SCHOOL OF MEDICINE, NEW ORLEANS, LOUISIANA

**Bruno Vogt, MD**
**Daniel Pouly, PhD**
UNIVERSITY HOSPITAL OF BERN, BERN, SWITZERLAND

## Skin, Muscle and Bone

20 GRANTS

**NEW**
**Justine Bacchetta, MD, PhD**
**Irma Machuca-Gayet, PhD**
HOSPICES CIVILS DE LYON
UNIVERSITÉ DE LYON, LYON, FRANCE

**Robert Ballotti, PhD**
**Christine Chiaverini, MD, PhD**
FACULTÉ DE MÉDECINE, NICE, FRANCE

**Andrea Del Fattore, PhD**
**Giulia Battafarano, PhD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Paul Grimm, MD**
STANFORD UNIVERSITY SCHOOL OF MEDICINE, PALO ALTO, CALIFORNIA

**Dieter Haffner, MD**
**Malgorzata Szaroszyk, PhD**
HANNOVER MEDICAL SCHOOL, HANNOVER, GERMANY

**Mary Leonard, MD, MSCE**
STANFORD UNIVERSITY SCHOOL OF MEDICINE, PALO ALTO, CALIFORNIA

**Robert Mak, MD, PhD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

**Richard Reimer, MD**
**Jacinda Sampson, MD, PhD**
**Mary Leonard, MD, MSCE**
**Paul Grimm, MD**
**Trinh Tina Duong, MPT**
**Feliks Kogan, PhD**
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

**Reza Seyedsadjadi, MD**
**Florian Eichler, MD**
**Lee Rubin, PhD**
MASSACHUSETTS GENERAL HOSPITAL, BOSTON, MASSACHUSETTS

# THE IMPACT of CRF RESEARCH

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

## Cystine Measurement and Cysteamine Toxicity Study

10 GRANTS

**Bruce Barshop, MD, PhD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

**Shawn Davidson, PhD**
PRINCETON UNIVERSITY, PRINCETON, NEW JERSEY

**Thomas Jeitner, PhD**
NEW YORK MEDICAL COLLEGE, VALHALLA, NEW YORK

**Elena Levtchenko, MD, PhD**
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

## Genetic Analysis of Cystinosis

5 GRANTS

**Katy Freed, PhD**
TEXAS BIOMEDICAL RESEARCH INSTITUTE, SAN ANTONIO, TEXAS

**Sihoun Hahn, MD, PhD**
SEATTLE CHILDREN'S HOSPITAL, SEATTLE, WASHINGTON

**Elena Levtchenko, MD, PhD**
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

**Eric Moses, PhD**
TEXAS BIOMEDICAL RESEARCH INSTITUTE, SAN ANTONIO, TEXAS

**Minnie Sarwal, MD, PhD**
UNIVERSITY OF CALIFORNIA, SAN FRANCISCO, SAN FRANCISCO, CALIFORNIA

## Stem Cells and Gene Therapy: Bone Marrow Stem Cells, Induced Pluripotent Stem Cells, Gene Therapy and Gene Editing

34 GRANTS

**Stéphanie Cherqui, PhD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

**Alan Davidson, PhD**
THE UNIVERSITY OF AUCKLAND, GRAFTON, AUCKLAND, NEW ZEALAND

**Bruno Gasnier, PhD**
PARIS DESCARTES UNIVERSITY, PARIS, FRANCE

**Paul Goodyer, MD**
MONTRÉAL CHILDREN'S HOSPITAL, MONTRÉAL, QUEBEC, CANADA

**Patrick Harrison, PhD**
UNIVERSITY COLLEGE CORK, CORK, IRELAND

**Vasiliki Kalatzis, PhD**
INSTITUTE OF MOLECULAR GENETICS OF MONTPELLIER, MONTPELLIER, FRANCE

**Winston Kao, PhD**
**Hassane Amlal, PhD**
UNIVERSITY OF CINCINNATI, CINCINNATI, OHIO

**Daniel Salomon, MD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Holger Willenbring, MD**
UNIVERSITY OF CALIFORNIA, SAN FRANCISCO, SAN FRANCISCO, CALIFORNIA

## Eye-Corneal Cystinosis Research

*11 GRANTS*

**Ghanashyam Acharya, PhD**
BAYLOR COLLEGE OF MEDICINE,
HOUSTON, TEXAS

**Stéphanie Cherqui, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**NEW**
**Morgan DiLeo, PhD**
**Xin Fan, PhD**
UNIVERSITY OF PITTSBURGH
SCHOOL OF MEDICINE,
PITTSBURGH, PENNSYLVANIA

**Morgan Fedorchak, PhD**
**Kanwal Nischal, MD, FRCO**
UNIVERSITY OF PITTSBURGH
SCHOOL OF MEDICINE,
PITTSBURGH, PENNSYLVANIA

**Jennifer Simpson, MD**
UNIVERSITY OF CALIFORNIA,
IRVINE,
IRVINE, CALIFORNIA

**Kang Zhang, MD, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

## Neurological

*17 GRANTS*

**Angela Ballantyne, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**Miriam Britt Sach, MD, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**Rita Ceponiene, MD, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**Florian Eichler, MD**
MASSACHUSETTS
GENERAL HOSPITAL,
BOSTON, MASSACHUSETTS

**Pascal Laforêt, MD, PhD**
**Hélène Prigent, MD, PhD**
RAYMOND POINCARÉ
UNIVERSITY HOSPITAL,
GARCHES, FRANCE

**Sophie Molholm, PhD**
**John Foxe, PhD**
ALBERT EINSTEIN COLLEGE
OF MEDICINE,
BRONX, NEW YORK

**Aude Servais, MD, PhD**
NECKER HOSPITAL,
PARIS, FRANCE

**Amy Spilkin, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**Doris Trauner, MD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

## Kidney Research

*25 GRANTS*

**Robert Chevalier, MD**
UNIVERSITY OF VIRGINIA,
CHARLOTTESVILLE, VIRGINIA

**Pierre Courtoy, MD, PhD**
**Christophe Pierreux, PhD**
DE DUVE INSTITUTE, LOUVAIN
UNIVERSITY MEDICAL SCHOOL,
BRUSSELS, BELGIUM

**Olivier Devuyst, MD, PhD**
UNIVERSITY OF ZÜRICH,
INSTITUTE OF PHYSIOLOGY,
ZÜRICH, SWITZERLAND

**Olivier Devuyst, MD, PhD**
**Marine Berquez, PhD**
UNIVERSITY OF ZÜRICH,
ZÜRICH, SWITZERLAND

**Allison Eddy, MD**
BC CHILDREN'S HOSPITAL,
VANCOUVER, CANADA

**Francesco Emma, MD**
**Anna Taranta, PhD**
BAMBINO GESÙ
CHILDREN'S HOSPITAL,
ROME, ITALY

**Benjamin Freedman, PhD**
UNIVERSITY OF WASHINGTON,
SEATTLE, WASHINGTON

**Elena Levtchenko, MD, PhD**
UNIVERSITY HOSPITAL,
LEUVEN, BELGIUM

**Robert Mak, MD, PhD**
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

**Tara McMorrow, MD**
UNIVERSITY COLLEGE
DUBLIN, BELFIELD,
DUBLIN, IRELAND

**Philip Newsholme, PhD**
CURTIN UNIVERSITY,
PERTH, WESTERN AUSTRALIA

**Daryl Okamura, MD**
SEATTLE CHILDREN'S
RESEARCH INSTITUTE,
SEATTLE, WASHINGTON

**Laura Rita Rega, PhD**
BAMBINO GESÙ CHILDREN'S
HOSPITAL, ROME, ITALY

**Mary Taub, PhD**
UNIVERSITY AT BUFFALO,
THE STATE UNIVERSITY
OF NEW YORK,
BUFFALO, NEW YORK

## Thyroid

*1 GRANT*

**Pierre Courtoy, MD, PhD**
DE DUVE INSTITUTE, LOUVAIN UNIVERSITY MEDICAL SCHOOL, BRUSSELS, BELGIUM

## Rat Model for Cystinosis

*3 GRANTS*

**Francesco Emma, MD**
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

**Olivier Devuyst, MD, PhD**
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

# THE IMPACT *of* CRF RESEARCH

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

## Molecular Study of Cystinosis in the Yeast Model

*3 GRANTS*

**Bruno André, PhD**
UNIVERSITÉ LIBRE DE BRUXELLES, GOSSELIES, BELGIUM

**Anand Bachhawat, PhD**
IISER MOHALI, MANAULI, PUNJAB, INDIA

**David Pearce, PhD**
UNIVERSITY OF ROCHESTER MEDICAL CENTER, ROCHESTER, NEW YORK

## Lab Equipment for Cystinosis

*9 GRANTS*

**Ghanashyam Acharya, PhD**
BAYLOR COLLEGE OF MEDICINE, HOUSTON, TEXAS

**Corinne Antignac, MD, PhD**
IMAGINE INSTITUTE, PARIS, FRANCE

**Bruce Barshop, MD, PhD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

**Sergio Catz, PhD**
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**Stéphanie Cherqui, PhD**
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

# 2022 CRF RESEARCH GRANT AWARDS

## FALL 2022 GRANT AWARDS $321,000

## $1,608,127

**Justine Bacchetta, MD, PhD and Irma Machuca-Gayet, PhD**
Hospital Femme Mère Infant, Bron Cedex, France
"The 2022 CYSTEA-BONE Project"
$246,000 TWO-YEAR STUDY

**Sergio Catz, PhD** *(Mentor)* **and Danni Chen, PhD** *(Fellow)*
The Scripps Research Institute, La Jolla, California
"Novel mechanistic and translational studies of inflammation in cystinosis"
$150,000 TWO-YEAR FELLOWSHIP

**Sergio Catz, PhD** *(Mentor)* **and Aparna Shukla, PhD** *(Fellow)*
The Scripps Research Institute, La Jolla, California
"Translational approaches to repair chaperone mediated autophagy in cystinosis"
$150,000 TWO-YEAR FELLOWSHIP

**Olivier Devuyst, MD, PhD** *(Mentor)* **and Marine Berquez, PhD** *(Fellow)*
University of Zürich, Switzerland
"Role of nutrient sensing and mTORC1 signaling in cystinosis"
$150,000 TWO-YEAR FELLOWSHIP

**Morgan DiLeo, PhD** *(Mentor)* **and Xin Fan, PhD** *(Fellow)*
University of Pittsburgh School of Medicine, Pittsburgh, Pennsylvania
"Liposome cysteamine carriers for eye drop formulation with long-term stability and smart release ability"
$75,000 ONE-YEAR FELLOWSHIP

**Francesco Emma, MD and Anna Taranta, PhD**
Bambino Gesù Children's Hospital, Rome, Italy
"Impact of diet composition on renal function and bone disease of Ctns-/- mice"
$287,320 TWO-YEAR STUDY

**Liang Feng, PhD**
Stanford University, Stanford, California
"Investigating the molecular basis of protein dynamics in cystinosis"
$245,000 TWO-YEAR STUDY

**Dieter Haffner, MD** *(Mentor)* **and Malgorzata Szaroszyk, PhD** *(Fellow)*
Hannover Medical School, Germany
"Musclin as a new marker for muscle health in cystinosis"
$74,915 ONE-YEAR FELLOWSHIP

**Jennifer Hollywood, PhD and Alan Davidson, PhD**
University of Auckland, New Zealand
**Herbie Newell, PhD** *(Collaborator)*
University of Sunderland, United Kingdom
"Evaluation of a novel drug combination treatment of CF10 and everolimus for nephropathic cystinosis in a new cystinotic rat model"
$229,892 TWO-YEAR STUDY

SEE LAY ABSTRACTS STARTING ON NEXT PAGE

# Pathophysiology of bone disease in nephropathic cystinosis: the 2022 CYSTEABONE project

**Justine Bacchetta, MD, PhD**
**Irma Machuca-Gayet, PhD**
HOSPITAL FEMME MÈRE INFANT, BRON CEDEX, FRANCE

## OBJECTIVE/RATIONALE:

Bone impairment has been recently described in patients with nephropathic cystinosis, with international recommendations for diagnosis and management published in 2019; the concept of "cystinosis metabolic bone disease" (CMBD) is now well established. Even though its exact pathophysiology remains unclear, this complication has a significant impact on patients' quality of life, because of an increased frequency of bone pains, deformations and fractures occurring in late teenage and early adulthood. It is therefore of utmost importance to understand the underlying mechanisms of such bone impairment.

## PROJECT DESCRIPTION:

We hypothesized that deep analyses of each bone cell type in cystinosis (i.e., osteoblast and osteoclasts) will allow to identify the "signature" molecular pathways of CMBD but also to provide new target gene candidates to design new therapeutical approaches. We have three main objectives at this stage of the project, based on our previous results obtained with 2 previous CRF grants:

1/ to finalize the experimental part of the inflammation/interleukin 1 project, so as to obtain a strong rationale to further propose a proof-of-concept trial in patients,

2/ to explore other regulatory pathways involved in CMBD based on our analysis of the ostoblastic transcriptome, and mainly BMP and Wnt signaling,

3/ to identify new targets to improve bone health in cystinosis using high-throughput pharmacological screening.

## RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

If our hypothesis of a deregulation of the interleukin 1 pathway in bone in nephropathic cystinosis is proved to be true, we will be able to propose new therapeutic perspectives such as therapeutic modulation of the interleukin 1 pathway, especially since this pathway was also recently identified as a potential target to attenuate cachexia in muscle cells by another team. On the long term, our results may provide a strong rationale for a clinical trial in the field; drugs targeting the interleukin 1 pathway already exist for inflammatory diseases.

## ANTICIPATED OUTCOME:

The CYSTEABONE-2022 project aims to better understand the underlying mechanisms of bone impairment, and to identify novel therapeutic approaches to improve (or prevent the onset of) bone symptoms in cystinosis. We are now part of the multi-center French and European RADICO cohort, that will definitively allow us to better characterize bone and mineral disease in "real-life" patients. The expected results that would be the most relevant for patients would be the confirmation of abnormal inflammation pathways in CMBD, that may lead us to further propose specific clinical trials.

# Liposome cysteamine carriers for eye drop formulation with long-term stability and smart release ability

**Morgan DiLeo, PhD,** *Mentor*
**Xin Fan, PhD,** *Fellow*
UNIVERSITY OF PITTSBURGH SCHOOL OF MEDICINE, PITTSBURGH, PENNSYLVANIA

## OBJECTIVE/RATIONALE:

The objective of this research is to develop a novel ocular drug delivery system for cystinosis patients with corneal crystals. Early initiation and good adherence to topical cysteamine are critical for ocular cystinosis treatment. However, the strict dosing regimen and poor stability of topical cysteamine are inconvenient and add to the burden of therapy, which make adherence difficult for many patients. Here we propose a novel eye drop formulation with smart drug carriers for improving cysteamine stability and reducing dosing frequency.

## PROJECT DESCRIPTION:

We want to provide initial proof of concept that our topical cysteamine formulation could maintain long-term cysteamine stability and provide sustained drug release with reduced dosage frequency to improve patient adherence. We will first demonstrate the inhibition effect of our formulation on the oxidation of cysteamine, this oxidation inhibition effect will contribute to the improved stability of cysteamine. Longer-term cysteamine stability in our drug delivery system will be studied. Next, in vitro cell studies and drug release studies will be used to evaluate the bioadhesive property and smart release ability of our formulation, which are key factors to maintain high drug stability during storage, but trigger release of cysteamine once applied to the eye.

## RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Our eye drop formulation with smart drug carriers would overcome the limitations of currently ocular cystinosis treatment with topical application of cysteamine. The main disadvantage is the extremely frequent administration requirement. In addition, cysteamine is highly susceptible to oxidative degradation to lose the therapeutic effect. Our goal is to develop an eye drop formulation of cysteamine that can maintain cysteamine stability over 30 days and reduce dosage frequency to once per day.

## ANTICIPATED OUTCOME:

We hope to develop an eye drop formulation that addresses the barriers to drug stability and patient adherence. We anticipate our eye drop formulation will maintain at least 90% stability of cysteamine for 30 days at room temperature. We also expect the eye drop will be safe, show high adhesion to mucus-producing cell, and achieve in vitro cysteamine release within the therapeutic window for 24 hours.

# CALL FOR SPRING 2023 GRANT APPLICATIONS

JOIN OUR RESEARCH LEGACY

When Nancy and Jeff Stack established the Cystinosis Research Foundation (CRF) in 2003, they were committed to aggressively funding cystinosis research to ensure the development of new and improved therapies and a cure for cystinosis. But never in their wildest dreams could they have imagined what has been accomplished in 20 short years. Since its inception, CRF has funded 223 multi-year research studies in 12 countries. Our researchers have published 103 articles in prestigious journals as a result of CRF funding. Every dollar donated goes directly to support cystinosis research.

The goal of CRF is to accelerate promising cystinosis research toward clinical trials. To that end, CRF prioritizes research that will lead to better treatments and a cure for cystinosis. CRF issues grants for bench, clinical and translational research, with a strong emphasis on translational and clinical research. CRF is interested in supporting new investigators and encourages them to apply either as research fellows or investigators.

In March, CRF announced $2.5 million was available for the Spring 2023 call for research and fellowship applications. The new grant awards will be announced in July.

In 2022, CRF issued nine new grants totaling $1,608,127 that will bring us closer to better treatments and a cure. All research applications received by CRF are evaluated by CRF's Scientific Review Board (SRB), composed of the leading international experts in the field of cystinosis. The SRB provides independent, objective reviews and recommendations for each research proposal submitted based on the NIH scale of standards. Additionally, the SRB follows grant review guidelines established by CRF and advises the foundation on the scientific merits of each proposal. CRF has created a thriving and collaborative international research community. If you are a scientist or researcher and would like to apply for a grant, please visit our website for more details.

Cure Cystinosis
International Registry

In 2021, CRF updated the registry questionnaire to include questions that are relevant to recent scientific advancements, new medications and patient care. CRF partnered with CoRDS *(Coordination of Rare Diseases at Sanford)* to create a new Cure Cystinosis International Registry (CCIR), the only international cystinosis patient registry in the world. The site includes a professional Research Portal so that researchers and scientists who register can access and view de-identified, aggregate cystinosis patient information. The registry will connect all of the stakeholders in the cystinosis community – the scientists, researchers, clinicians, pharmaceutical companies, patients and families – and provide them with resources that have never been available in one place before, all to accelerate patient care.

The value of the registry will only truly be realized with patient participation. We strongly encourage patients or caregivers to enroll in the registry and help identify the needs of patients with cystinosis. This information is essential to advancing cystinosis research.

Visit the CRF website to learn more about CCIR and enroll.

*WWW.CYSTINOSISRESEARCH.ORG/CURE-CYSTINOSIS-INTERNATIONAL-REGISTRY*

**WWW.CYSTINOSISRESEARCH.ORG/APPLY-FOR-RESEARCH-GRANT**

CRF is excited about the future of cystinosis research and is grateful to researchers for their interest in the cystinosis community. We look forward to working together to find better treatments and a cure for cystinosis.

CYSTINOSIS RESEARCH FOUNDATION

# SCIENTIFIC REVIEW BOARD

The CRF Scientific Review Board (SRB) is composed of leading cystinosis scientists, researchers, and clinicians from around the world. We are indebted to our Scientific Review Board members for their leadership, guidance and commitment to improving the lives of adults and children with cystinosis. THANK YOU!

## CHAIR

Corinne Antignac, MD, PhD
*Professor*
Laboratory of Hereditary Kidney Diseases,
Imagine Institute (Inserm U1163)
PARIS, FRANCE

## BOARD MEMBERS

**Sergio D. Catz, PhD**
*Professor*
Department of Molecular Medicine
The Scripps Research Institute
LA JOLLA, CALIFORNIA

**Stéphanie Cherqui, PhD**
*Professor*
Department of Pediatrics
Division of Genetics
University of California, San Diego
LA JOLLA, CALIFORNIA

**Francesco Emma, MD**
*Head of Pediatric Nephrology*
*Director of Nephrology Laboratory*
Bambino Gesù Children's Hospital
ROME, ITALY

**Larry Greenbaum, MD, PhD, FAAP**
*Division Director of Pediatric Nephrology*
*Bernard Marcus Professor of Pediatric Nephrology,*
Emory School of Medicine
*Executive Clinical Director*
Children's Healthcare of Atlanta
ATLANTA, GEORGIA

**Paul C. Grimm, MD**
*Professor of Pediatrics*
Pediatric Nephrology
Stanford University School of Medicine
PALO ALTO, CALIFORNIA

**Julie R. Ingelfinger, MD**
*Deputy Editor of the*
*New England Journal of Medicine*
*Professor of Pediatrics*
Harvard Medical School
BOSTON, MASSACHUSETTS

**Stephen L. Jenkins, MD**
*Assistant Professor, Hospitalist*
Department of Internal Medicine
University of Utah Hospital
SALT LAKE CITY, UTAH

**Aude Servais, MD, PhD**
*Senior Nephrologist,*
*Department of Adult Nephrology*
Necker Hospital, Paris Descartes University
PARIS, FRANCE

**THANK YOU FOR YOUR DEDICATION TO THE GLOBAL CYSTINOSIS COMMUNITY**

# CYSTINOSIS MAGAZINE IS

*going green!*

Starting in Spring 2024, the Cystinosis Magazine will be making the long-awaited transition from print to digital. With deforestation continuing to destroy ecosystems across the planet, every effort – big and small – can help make a difference.

As a foundation dedicated to making the world a better, healthier place, we are excited to officially bring the Cystinosis Magazine into the digital space and do our part for the environment. *Plus,* you'll enjoy the convenience of having our magazine available at your fingertips, whenever and wherever you are!

**Don't miss out on any future (or past) editions! Sign up for our mailing list for the digital Cystinosis Magazine today by scanning this QR code and filling out our contact form!**

cystinosis RESEARCH FOUNDATION

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President
*Cystinosis Research Foundation*

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*Boingo Wireless*

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Sr. Technical Director
*Princeton Hydro, LLC*
*Hope for Brooke*

**Jill Emerson, CPA**
*Hope for Brooke*

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*Tina's Hope For A Cure*

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University of Utah Hospital
*Sam's Hope for a Cure*

**Angela Kirchhof**
*Hope for Hayden*

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Chief Administrative Officer
*Village of Consort, Alberta, Canada*

**David W. Mossman**
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cystinosis RESEARCH FOUNDATION

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## MISSION

*The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised over $67 million with 100% of your donations going to support cystinosis research.*

## EDUCATION

*CRF is dedicated to educating the medical and public communities about cystinosis to ensure early diagnosis and proper treatment.*

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TOGETHER

Strong roots
produce strong trees.
We grow stronger
in community.

The tree rings
reveal our history
as we all grow
in the CRF story.

Our dedication.
Our connection.
Our cystinosis journey
**TOGETHER**
through the years.

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