cystinosis magazine

For friends and supporters of the Cystinosis Research Foundation

GROWING TOGETHER

SPRING 2018

GROWING TOGETHER ISSUE

GROWING TOGETHER

Through the Cystinosis Research Foundation, families, patients, doctors, and researchers from all over the world come together and share stories, information, and lean on each other for hope, strength, and inspiration. This issue includes stories of growth and persistence from all sides in pursuit for better treatments and a cure. We are all on this journey growing together!

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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Art Direction and Printing: Idea Hall

Emma Suetta with her cystinosis champion, Christy Smiley.

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CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

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 $40 million for cystinosis research in an effort to find a cure.

WELCOME

CRF CHAMPIONS

CRF COMMUNITY

RESEARCH HIGHLIGHTS

ANNOUNCEMENTS

FAMILY STORIES

CYSTINOSIS MAGAZINE IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

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DEAR FAMILY AND FRIENDS

We are grateful to all of you who have grown together with us as we blaze the path to find better treatments and a cure for cystinosis. We have come so far since 2003 when Natalie scribbled her birthday wish on a napkin, "*to have my disease go away forever.*"

Her wish was the beginning of this story, but each step taken since her wish has been taken hand-in-hand with you. Nothing could have prepared us for the outpouring of love, support, kindness and commitment from you, our stalwart community of family and friends who have embraced our journey and grown with us as we move closer to the cure.

We love this time of the year — it is a season of hope and renewal. We started the year off with our 6th International Research Symposium where CRF funded researchers gathered to discuss their research projects and to learn from each other. The symposium concluded with several researchers agreeing to collaborate in an effort to accelerate the research process. We have come so far; we have built a strong foundation for innovative research which continues to flourish and produce extraordinary results. We have a wonderful article about the symposium on page 29.

Every year we look forward to the Day of Hope family conference held in April this year. Over 300 people attended the conference – it was a celebration of our cystinosis community and our researchers who have dedicated their careers to helping our community (page 18). We are certain you will enjoy the stories in this magazine about the Champions in our community who surround us with their passion for our cause. You will read the awe-inspiring stories of our families and adults with cystinosis — their courage and determination are an example of how to live life in the face of so many challenges.

We are thankful for you every day because you have embraced our cause and grown with us while we have expanded our research efforts and flourished as a community. As Margaret Mead so aptly stated, "Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has." We are changing lives together, you are giving hope to thousands and possibly even millions who will benefit from our research discoveries. We are closer to a cure than ever before because of your commitment to our community.

RESEARCH FRONT AND CENTER

As you know, cystinosis damages every cell in the body so because of that, our research portfolio is comprehensive. We have focused our efforts on areas that cystinosis affects first and most severely – the kidneys and the eyes — but have also targeted research that seeks answers on how to treat muscle wasting, neurological issues and bone abnormalities caused by cystinosis.

We seek answers to the pathogenesis of cystinosis, by funding researchers who study the basics of the cell so that we better understand the "why" of cystinosis. We fund researchers who are focused on various proteins in cystinosis, molecules to enhance and improve treatments, and autophagy which is a metabolic process in the body that destroys cells.

We have important multiyear studies focused on newborn screening, improved cysteamine treatments, novel deliveries for corneal cystinosis, iPS cells and studies to screen already approved drugs to possibly treat cystinosis. The breadth of the research is vast but our focus is narrow; every study we fund will help us move closer to better treatments and a cure.

In order to find a cure and better treatments for a complicated disease like cystinosis, our strategy involves funding bench, clinical and translational research. We keep all of your donations working throughout the year by funding new grants biannually. Since 2003, CRF has funded 164 multi-year research studies in 12 countries. Our researchers have published 72 articles in prestigious journals as a result of CRF funding. Today, CRF is the largest private fund provider of cystinosis research in the world.

We have created a synergistic research community that has made significant discoveries and because our research teams work collaboratively, the discoveries made by one group of researchers invariably helps the work of other research labs. We expect that there will be more important discoveries and enhanced collaboration as we continue to grow our science community.

We are thankful for the dedication of the science and medical communities who work every day to solve the mysteries of cystinosis and who treat our children as if they were their own. With you by our side, we have built a strong foundation for innovative research which continues to flourish and produce extraordinary results.

From the beginning, all CRF operating costs have been privately underwritten so that 100 percent of your donations to CRF goes towards research.

THIRTEEN NEW STUDIES IN FIVE COUNTRIES TOTALING OVER $2,464,533 FUNDED IN 2017

We are pleased to announce that in 2017 we issued 13 new grants totaling over $2.46 million in research awards. The grants were awarded to researchers in the United States, Canada, France, Italy, and Switzerland. The grant recipients for fall 2017 are listed on page 64 along with a lay abstract of their study listed on page 75. The new grants focus on stem cell research, distal myopathy, novel treatment therapy, newborn screening and research focused on the cellular and molecular aspects of cystinosis.

STEM CELL AND GENE THERAPY – POTENTIAL BEYOND CYSTINOSIS

Dr. Stéphanie Cherqui at University of California San Diego (UCSD) continues to work every day using stem cells and gene therapy. With a one-time treatment in mice, Dr. Cherqui reversed cystinosis including corneal cystinosis, thyroid dysfunction and kidney disease. Dr. Cherqui's treatment for cystinosis will involve using the patients' own stem cells and gene-correct them to introduce a functional CTNS gene before transplanting them back in the patients. This approach, called autologous transplantation, is safer than using foreign stem cells, but requires optimization of the gene-correction step.

Based on Dr. Cherqui's findings on the impact of hematopoietic stem cell transplantation in cystinosis and the mechanism of action, she successfully treated two other disorders in the mouse models to date: Friedreich's Ataxia, a fatal neuromuscular degenerative disorder and Danon disease, a lysosomal disorder that results in cardiac disease in early adolescence requiring heart transplantation.

Dr. Olivier Devuyst a CRF funded researcher in Switzerland, demonstrated that Dr. Cherqui's hematopoietic stem cell transplantation could rescue the mouse model of Dent disease, a chronic kidney disorder and patients require kidney transplantation.

Dr. Cherqui expects to file the IND (Investigational New Drug) application very soon. We are optimistic that the FDA will give their approval to begin a phase 1 clinical trial to treat the first patients with Dr. Cherqui's promising treatment before the end of the year.

NOVEL TREATMENTS FOR CORNEAL CYSTINOSIS:

Corneal cystinosis is the build-up of cystine crystals in the eyes that causes photophobia (extreme sensitivity to light) severe eye pain, and sometimes, blindness. There is an existing eye drop treatment but it is rigorous (the drops must be taken every waking hour) and painful for many patients. The demanding protocol of hourly eye drops results in poor

compliance. Currently we are funding two researchers focused on novel treatments for corneal cystinosis. We are funding Ghanshyam Acharya, PhD at Baylor College of Medicine whose study involves nano technology to deliver medication to the eye. He developed a nanowafer that can be loaded with medication, placed in the eye where it can deliver medication potentially over several hours. We are also funding Morgan Fedorchak, PhD at the University of Pittsburgh. She had developed a hydrogel drop that is filled with small microspheres that are loaded with medication. She is hopeful that the drop will only need to be taken once a day but will have the potential to deliver an entire day's worth of medication. These two novel treatments are promising; we hope they will result in clinical trials in the near future.

CLINICAL TRIALS YIELD IMPORTANT INFORMATION

CRF has funded many clinical trials over the years. The trials we have supported that are currently open include studies for novel drug treatments, distal myopathy, sleep disturbance, bone abnormalities, quality of life studies and cystine measurements. These clinical trials yield important data that can be translated into potential new treatments that will improve life with cystinosis. Your support has enabled us to translate the information gleaned from these studies to potential new treatments and a possibly a cure!

CRF FUNDED RESEARCH WILL HELP OTHER DISEASES

In addition to CRF funded stem cell and gene therapy work helping others, many more discoveries made by CRF researchers are currently being applied to other more prevalent and well-known disorders and diseases including other corneal diseases, kidney diseases and genetic and systemic diseases similar to cystinosis. Your support of cystinosis research has reached far beyond the cystinosis community. A cure for cystinosis will help find cures for other diseases, potentially helping millions of people.

WE WILL CHANGE THE CYSTINOSIS WORLD TOGETHER

We remain committed and focused on funding research that will help our children and save their lives. We pray for a life without medications, without pain, without muscle weakness, without emergency room visits, blood draws and gtubes. We do not want our children to worry about their life expectancy.

Your unwavering support, your commitment to research and your compassion and love for our community gives us hope. We have made extraordinary progress and with your continued support, generosity and love, we will soar to new heights and find a cure for cystinosis.

Thank you for supporting cystinosis research, for standing by our side and for embracing our community. We are blessed by your encouragement, your prayers and your steadfast belief that together we will find the cure. We are humbled by your dedication to the cystinosis cause. Without you, we simply would not be where we are today.

With heartfelt thanks and gratitude,

Nancy & Jeff

WE'RE GROWING TOGETHER

A LETTER FROM NATALIE

Dear Family and Friends,

It has been one year since I started my job at Hope Builders. I enjoy having a career in the nonprofit field; I have gained so many skills and have had so many new experiences and love working for an organization that impacts the Orange County community in such a meaningful way.

Although I enjoy my new career, it has been quite a challenge adjusting to the work schedule. As most of you know, this is my first full-time job since finishing graduate school. While I was a student, I would typically sleep in and schedule my classes in the afternoon. I am not a morning person! With a job, I need to adjust to an early start time which has been really hard for me. I blame the challenges I face on cystinosis.

The medicine I have to take can be unbearable at times, with side effects ranging from extreme fatigue, a strong sulfur odor, red puffy eyes, to a throbbing headache. When I first started my job, it took me a solid three months to fully adjust to my new schedule. My mom didn't think I was going to be able to maintain a full-time job because she saw me struggle each day, extremely fatigued after work with little to no energy. I would take a long nap every day after coming home from work and each morning I struggled to get out of bed. Cystinosis is an extremely strenuous disease on the body, and many people do not understand the list of complications that result from this terrible disease.

Though having a full-time job takes a lot out of me, I am proud of myself for being able to show up to work every day and challenge myself both professionally and personally. I want to continue to follow my dreams and pursue my career in social services. I want to make a real difference in the lives of others. I want to give back to the community because it has done so much for me. Cystinosis is just a small part of who I am. It will not stop me from pursuing my dreams and living my life to the fullest.

Thank you to everyone who has supported me throughout my journey. Thank you to my family, my friends, the researchers and doctors, and most of all, the cystinosis community. I am so incredibly blessed to be surrounded by such strong, resilient people. Together, we can do anything.

With Love,

Natalie Stack

Cystinosis is just a small part of who I am. It will not stop me from pursing my dreams and living my life to the fullest.

WE **CELEBRATE** OUR **CRF COMMUNITY** AND ARE GRATEFUL EVERY DAY FOR YOUR SUPPORT!

CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

100% of your donations directly support cystinosis research

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. Build-up 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 500 people, mostly children, in North America, and about 2,000 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.***

The Cystinosis Research Foundation was established in 2003 with the sole purpose of raising funds to find better treatments and ultimately a cure for cystinosis.

Today, CRF IS THE LARGEST FUND PROVIDER OF GRANTS FOR CYSTINOSIS RESEARCH IN THE WORLD, funding more than 164 studies in 12 countries.

CRF has raised over $40 million with 100% of all 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.

CRF was founded in 2003 after Natalie Stack made a wish on the eve of her 12th birthday.

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

We are on this journey together growing closer to the cure with more new clinical trials than ever before. It is because of you that CRF has been able to fund extraordinary researchers across the globe.

IN 2017, CRF FUNDED:

13 research grants

TOTALING

$2.46 million

72 articles published
based on CRF-funded research

CRF is the largest fund provider of grants for cystinosis research in the world

SINCE 2003, CRF HAS ISSUED:

164 research grants
in **12 countries**

CHARITY NAVIGATOR
★★★★
Four Star Charity

GROWING TOGETHER

A DREAM HOME FOR CHARLOTTE

THE RENOVATION OF THEIR FIXER-UPPER WAS BEYOND ANYTHING THE COE FAMILY COULD HAVE HOPED FOR. UNTIL CHUCK WOOD MADE IT HAPPEN.

By Dennis Arp

It all started with kitchen cabinets. Megan and Mike Coe bought their Webster, New Hampshire, home as a fixer-upper, knowing that it would need considerable renovation, which they planned to do systematically, over time. That was before their then-6-month-old daughter, Charlotte, was diagnosed with cystinosis. Suddenly home repairs became secondary compared with meeting Charlotte's constant care needs. Still, those cabinets.

"They were falling apart, to the point that sawdust was spreading on the countertops" recalls construction contractor Chuck Wood, who was summoned for an assessment. "They needed a more-sterile area to prepare Charlotte's meals to be sure she was getting what her body needs. Something had to be done."

Wood, founder of Colonial Builders, had never met the Coes before he was called in May 2017 by Amanda and Eric McKim, longtime friends of the Coes.

"Eric talked about the kitchen cabinets, and he explained about Charlotte's illness. It became an emotional moment for both of us as he explained what the ailment is and how challenging it is for Charlotte and the whole family," Wood relates. "I said, 'I'm in — whatever they need.'"

From the outset of his initial walk-through, Wood realized that the cabinets were just the beginning.

"They really needed a whole new kitchen," he says. "On the way in, I saw that the roof wasn't in great shape. The siding wasn't good. The home originally was a three-season camp, so there were issues with insulation. I saw lots of different issues — the laundry room, the mudroom, doors, the flooring, you name it."

Wood started making a list. And before he left the Coe family's home, he sat down with Mike and Megan to get to know them better. He wanted them to feel good about their home and what it could become.

"They're amazing people — extremely humble," Wood says. "I wanted them to almost dream about what they would like to see in their home, to think beyond the limitations of resources."

Afterward, during the drive home, Wood developed a dream of his own.

"It was a rough ride home for me, sitting in the truck, thinking about Charlotte and the family and the pain they must be going through," Wood says. "They couldn't take care of these things because everything was going toward Charlotte, which is the way it should be. I made a decision: I'm going to fix their home, and I don't care what it takes. When I got home, I explained the situation to my wife, and she said, 'Whatever you want to do, I'm 100 percent behind you.'"

Wood started reaching out to his subcontractors as well as to other industry friends and associates. The story of the Coes' needs and Wood's passion for the project won allies by the score. His regular roofing contractor donated materials and labor, and his plumber did the same. The McKims started a crowdfunding campaign called Charlotte's Village on GoFundMe, and donations started pouring in.

"Once I got the logistics of things situated, I did up some paperwork for Mike and Megan – basically an estimate," Wood says. "I made an appointment and then went back to their house. I said, 'Let's go for a walk.'"

CHARLOTTE COE
Webster, New Hampshire

They retraced their original walk-through as Wood outlined his plans. Eventually the three of them ended up at the couch, where Wood shared his detailed project list, with his cost estimate at the bottom. It took a moment for Megan and Mike to comprehend what they were reading.

"He said the cost would be zero dollars," Megan recalls. The Coes' gratitude overflowed.

"We were speechless," Megan says. After the initial shock, the Coes got one more surprise from Wood. Because his renovation plans were comprehensive, he knew that they couldn't live in the house during the work. He had talked to a client who owns a nearby vacation home, and the Coes were invited to stay there during the two weeks of intensive activity.

"Things came together unbelievably well," Wood says. "The amount of support we got from friends and people in the community — it just kept coming."

During the project itself, an amazing array of community members contributed. Many made sure the work crews were fed, from pastries in the morning to pizza and subs at lunch. Skilled contributors included general carpenters and a stone mason, who added a façade that transformed a cinderblock chimney into a work of art. When the indoor work was done, a local nursery sent a team to clean up the yard and plant flowers.

"The fantastic people I met on this project will be friends for life," Wood says. "I really don't know how to explain it. I love what I do, but this project was extra special. I woke up with the biggest smile on my face, and at the end of a long day, I still had a smile on my face. I've done pro-bono work before, but never like this, where everyone had the same mentality. It was awesome."

The project required a special culmination, and Wood was ready with that as well. Mike and Megan were driven to a place just up the road from their home, where the McKims covered their friends' eyes and led them to the end of their driveway.

At the reveal, the Coes' emotional reaction was the capper for a project full of joy, Wood says.

"It was more than we could have even begun to hope for," Megan says.

About a month after the Coe family had settled back into their home, about 60 of the people who contributed to the project gathered to celebrate Charlotte's second birthday as well as the group's collective success. Folks brought food, cake, balloons and presents. Chuck Wood got a chance to chat and trade hugs with his bundle of inspiration.

"Charlotte is full of life," he says. "She has such an intelligence level; she's really linguistically gifted. But what really grabs me is how loving and caring she is. On a deep and emotional level, you can tell that she's genuinely interested in you. And she loves giving hugs."

These days, new construction projects sometimes take Wood down the road that passes the Coes' house. Texts get traded, and Wood receives an invitation to stop for a cup of hot cocoa. He doesn't have to be asked twice.

"We have a bond now," he says of his affection for the Coes. "They'll never get rid of me."

CYSTINOSIS COMMUNITY CHAMPIONS

CHRISTY SMILEY

FEEDING A FRIENDSHIP

CHRISTY SMILEY'S TRI-TIP FUNDRAISER HELPS SUSTAIN A CYSTINOSIS FAMILY THROUGH THE GENEROUS SPIRIT THAT BINDS HER COMMUNITY.

By Dennis Arp

As a mother of three in a logging family with roots at the northern edge of California, Christy Smiley knows all about the simple joys of small-town life.

"It amazes me what people will do when someone needs help," she says. "Everyone just pulls together to get things done. One person might have an idea, but you never have to do it alone."

Smiley didn't know the Suetta family all that well and she had never heard of cystinosis when she learned that then 2-year-old Emma Suetta had been diagnosed with the rare genetic disorder, forcing her to stay in a far-off hospital for 26 days. Smiley realized the Suetta family life had been turned on its head.

She had an idea. "I had done my share of tri-tip dinner fundraisers," Smiley says. "It's something I knew how to do."

Word went out, and an event to benefit the Suettas took shape, thanks to Smiley and other residents of Etna, California, population 700. Throughout the Scott Valley in Siskiyou County, folks rallied to Smiley's idea on a scale beyond anything they had tried before. By the end of the event, Smiley and her neighbors had prepared and handed out more than 700 tri-tip dinners, raising about $7,000 to offset the cost of Emma's care.

Two years later, Smiley remains an enduring champion for the Suetta family.

"Christy's support means the world to us," says Shelly Suetta, Emma's mom. "She's a giving person, always thinking of everyone else. She's just that type of person. If we didn't have the support we have from folks locally, I don't know how we could have gotten through everything."

It was June 2015 when Shelly and Derek Suetta received the life-changing news that Emma had cystinosis. After weeks of tests and uncertainty, they had been referred to a nephrologist at University of California San Francisco Medical Center, about a seven-hour drive from their home. Emma would spend nearly a month in the hospital, getting treatment to stabilize her health. Ever since, Emma, her parents, and her older sister Lillyannna have been adapting to new realities.

"We were devastated — in tears — as we figured out what to do," Shelly says, recalling the initial diagnosis and hospital stay. "We were lucky that our social worker knew of another cystinosis family, and we got connected to CRF (Cystinosis Research Foundation). With a rare disease, you feel so alone, but we quickly learned that we weren't alone."

After Emma's long hospital stay, the Suettas were greeted by new friends and resources, but also a new daily regimen of treatments and medications that is all too familiar to cystinosis families. Their vocabulary expanded to include terms like PICC line and G-tube. Plus, there were the bills – it was all more than a little overwhelming. Thank goodness they were also greeted by the love of their neighbors.

Strangers showed up with meals and gas cards arrived anonymously in the mail. Donors provided about a year's worth of diapers, recognizing the special needs of a toddler who drinks four liters of water a day.

Still, Smiley wanted to do more.

"When someone's in need, you want to do as much as you can to make it as easy as possible for them," Smiley says. "You want to give them something to look forward to."

She had led or participated in numerous

EMMA SUETTA
Etna, California

fundraisers, especially for local school causes and sports teams. When she announced the tri-tip dinner event for Emma, "lots of friends and others saw what we were doing and let me know on Facebook that they would help," Smiley says.

The folks at Dotty's restaurant on Highway 3 in Etna volunteered to cook the tri-tip, and Fort Jones Elementary School made its kitchen available for use. An army of others donated their skills and time to assemble packaged meals that included vegetables, salad, beans, and a roll, in addition to the tri-tip. Another generous volunteer set up a bake sale in the school parking lot. For $12, participants got a hearty take-home meal plus the warm feeling of knowing they had made a difference in Emma's life.

"With so much help, it all went smoothly," Smiley says. "Everyone in our little town is familiar with these dinners, and now I think that everyone in the valley knows what cystinosis is. It definitely brought awareness to our community."

The Suettas are certainly aware that they have a champion in Smiley.

"She has three kids of her own, and they're all friends of ours now," Shelly Suetta says. "She's involved in so many things. I don't know how she has the time."

Smiley returns the compliment.

"Shelly's a wonderful mom, and I can't imagine all that she has to do every day," Smiley relates. "She blends all of Emma's food for her feeding tube to make sure she gets all the nutrients she needs. They're a special family, and they're helping Emma thrive."

Indeed, the little girl who faced a mountain of uncertainties when she was first diagnosed recently celebrated two years of hospital freedom.

"The silver lining of being diagnosed so early is that she doesn't know of a life without a G-tube," Shelly says of Emma. "She's so happy, and so resilient. She'll be sick one moment, then say, 'I'm OK,' and run off to play. At 4, she's now able to throw up in the toilet. I told my husband the other day, you know you're a cystinosis parent when that's a milestone in your life."

No doubt there are more breakthroughs on the horizon as Emma continues to grow and meet her many challenges. Having friends like Christy Smiley providing love and support makes the passage that much richer, Shelly notes.

"She has really adopted us for this journey."

CYSTINOSIS COMMUNITY CHAMPIONS

PAUL DAMUDE

GREAT PUMPKINS

SEEKING A PARTNER FOR HER CREATIVE FUNDRAISER, ABBI MONAGHAN FINDS A CHAMPION IN FARMER PAUL DAMUDE, WHO ENSURES HER PROJECT HAS ROOM TO GROW.

By Dennis Arp

In the Niagara Region of Ontario, Canada, Paul Damude tends orchards and tills fields that have been in his family since 1786. Over that time, countless produce buyers and dealmakers have traveled Cataract Road to stop at the Damude farm. Still, the initial visit by Abbi Monaghan stands out from the rest.

Abbi was 10 when she stepped into the family's roadside store for the first time two years ago. She was with her parents, but it was Abbi who asked how much it would cost to buy all of the pumpkins — more than 100 — in the Damudes' fall harvest. Paul Damude remembers the moment well. "She is quite the cutie," he says.

Abbi's plans to paint the pumpkins and sell them to raise money for cystinosis research quickly sealed the deal. Damude refused to accept any money, and the Monaghan family drove away knowing that Abbi's fundraising idea was set for the season. Thanks to Damude, it has been set ever since. "She's a good negotiator," Damude says with a laugh.

More than a good neighbor, Damude has become a great champion for Abbi and the cause of cystinosis research, says Katie Monaghan, Abbi's mom.

"He is super generous," Katie enthuses. "He's always wanting to know more about cystinosis and about how Abbi is doing. He's onboard with anything that helps the cause. He does more and gives more every year."

For the past three seasons, Abbi and her family have been painting and selling pumpkins as a fundraiser during the run-up to Halloween. Abbi has lived with the effects and the many challenges of cystinosis since she was first diagnosed at 18 months old, but through it all she has been determined to support the Cystinosis Research Foundation (CRF) and its search for a cure. She found the perfect way with her pumpkin project.

"She loves art," Katie Monaghan says of Abbi. "She goes to art camp each summer, and she's always creating."

During the first season of planning, painting and otherwise decorating pumpkins for sale, the result was about 100 creations. The Monaghans and friends promoted the sale on social media (using the Facebook page and hashtag Abbi's Road) and by other means, including word-of-mouth in and around their hometown of St. Catherines. Though the family was unsure just how much public interest there would be, it turns out there was no need for concern. The pumpkins sold out in about a half-hour.

"Things have grown every year from there," Katie says. "Now each fall, you see the pumpkins on porches all over our neighborhood."

Last season, the painted-pumpkin inventory expanded to about 300, with Abbi still doing most of the creative work. A donation by Rustoleum of six cases of spray paint helped the cause greatly, but painting is just part of the decorating story. Abbi also bedazzles many of her creations with jewels and sparkles as she bolsters the range of her designs.

"It gets a little chaotic as our garage fills up and then the back patio fills up," Katie says. "All the decorating happens outside, but stuff gets tracked in. We're constantly vacuuming up sparkles."

ABBI MONAGHAN
St. Catharine's, Ontario, Canada

Among the top sellers in Abbi's product line are pumpkins of silver and gold, while others reflect a unicorn theme or are covered in faux jewels. A new addition was particularly popular last season.

"People wanted to pay double for a football pumpkin," Katie says.

General demand for Abbi's creations is so high that some customers start arriving on sale day well before the announced time. The most recent sale also included scarves tie-dyed by Abbi and bracelets made by a friend of the Monaghans, Teresa Koch. All told, the day netted $3,300 for the CRF and the cause of research.

"Thanks to Paul, all the proceeds from the sale go to improving lives," Katie says.

Damude relates that he gets far more satisfaction from making his annual donation than he ever did from selling his pumpkins at his farm store, where his main cash crop is apples.

"When you meet Abbi and learn what cystinosis patients go through, donating what you can is the least you can do," Damude says. "For sure you have to do your part."

Everyone involved is particularly excited about this fall's sale. Soon, Damude will begin his largest-ever pumpkin planting, with Abbi participating in every step of the process. Plans are that after this year's harvest, Abbi will do the decorating and selling right at Damude's farm, adding new possibilities for customer traffic.

Damude isn't yet sure where Abbi and her family will do the decorating. All he knows is that "we will find the room," he says.

"I can't wait for her to be out here and to help her learn all about planting and growing the pumpkins," Damude adds. "It's going to be a lot of fun for everyone."

Katie agrees.

"Because Abbi will have a hand in things from beginning to end, it will be a great learning and bonding experience, for her and for Paul," she says.

There's no doubt that for the Monaghans, Damude fits the definition of a champion.

"If you think there are fewer genuine people in the world these days, you definitely want to meet Paul," Katie says. "He's quiet, but he has a huge heart. And there's just no one out there who's more genuine than he is."

DR. LAVJAY BUTANI

Dr. Lavjay Butani "Dr. B."

JENNA AND PATRICK PARTINGTON'S DOCTOR

UNIVERSITY OF CALIFORNIA, DAVIS

CHILDREN'S SPECIALTY CLINIC

SACRAMENTO, CALIFORNIA

We first met Dr. Lavjay Butani when Jenna and Patrick were 16 months old. His care was recommended by a metabolic specialist following the twins' diagnosis of cystinosis. Since that time, our family has developed a deep affection for Dr. Butani. He is gentle and kind, caring and concerned, and he looks after us as a family.

Dr. Butani is a doctor of pediatric nephrology (kidney care) at the University of California, Davis, Children's Specialty Clinic in Sacramento, California. Because cystinosis most commonly affects kidney health first, a nephrologist almost always sits at the helm of cystinosis care, tending to kids' comprehensive medical needs as they relate to cystinosis and chronic kidney disease. Dr. Butani makes sure that Jenna and Patrick are followed by doctors in other specialties as needed, including ophthalmology, orthopedics, gastroenterology, endocrinology etc.

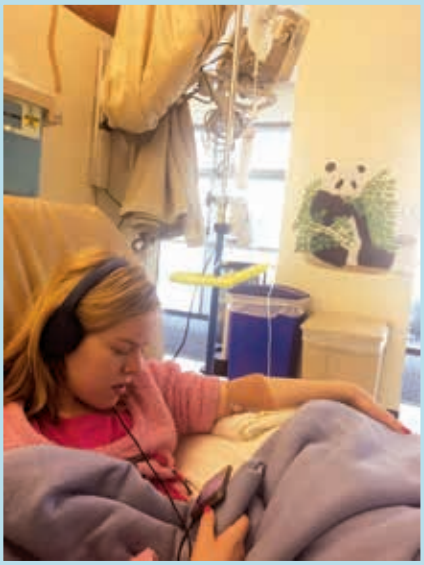
"Dr. B." sees the twins every three months, at which time they have a medical exam that includes reviewing their vitals: blood pressure, stature, and a physical exam. After each appointment, the kids have blood work done, which helps Dr. Butani determine how the kids' myriad medications are working, and he makes adjustments as needed.

When the twins were active babies and toddlers, the exam room where we would meet Dr. B was mayhem! Patrick and Jenna would bash toys on the wall that clicked and clacked, and scales and medical tools in the room were irresistible to them. I would often try in vain to recall the questions I had for Dr. Butani, as Kevin and I did our best to wrangle the kids. The visits were an exercise in patience – until Dr. Butani walked in with his calming, reassuring nature. Those days made it clear that Dr. B. is meant to work with children, as much as he is meant to serve in a position to reassure and offer guidance to their parents. He is unflappable.

With the exception of the blood draw, the kids now look forward to their visits with Dr. Butani, and they always leave with new knowledge about their bodies and about cystinosis. While the words of mom and dad about their self-care can seem nagging, redundant and worrisome, the words of Dr. Butani are thoughtfully considered and carefully employed by Jenna and Patrick. The difficult undertaking of managing cystinosis is broken down into understandable tasks that our 13-year-olds seem to be vested in. I still approach each of the kids' appointments with a checklist of questions for Dr. Butani; and each time we depart the Med Center, the boxes are checked and I am reassured that Jenna, Patrick, Kevin and I are doing all we can to stave off the devastating effects of cystinosis for as long as possible.

WHILE THE WORDS OF MOM AND DAD ABOUT THEIR SELF-CARE CAN SEEM NAGGING, REDUNDANT AND WORRISOME, **THE WORDS OF DR. BUTANI ARE THOUGHTFULLY CONSIDERED AND CAREFULLY EMPLOYED BY JENNA AND PATRICK.**

Most recently, Dr. Butani saw Jenna through a nearly six-week bout with mononucleosis. His care, in cooperation with the U.C. Davis Pediatric Infusion Center, made it possible for Jenna to get outpatient IV hydration when necessary, helping her trudge through her illness without hospitalization.



As always, I had direct access to Dr. Butani via email whenever we needed reassurance. In today's world of medicine, I know that is the exception, not the rule. Jenna and Patrick agree: It is difficult to imagine living with cystinosis without the dedicated care of Dr. Butani.

‘TINA, YOU HAVE A LETTER FROM YOUR DOCTOR’

Dr. Amira Al-Uzri

TINA FLERCHINGER’S DOCTOR

OREGON HEALTH & SCIENCE UNIVERSITY HOSPITAL

PORTLAND, OREGON

Dear Tina,

Most people have trepidation when they receive a letter from their doctor and think, “What could be in this letter?” You need not worry. This letter is different. It discusses my perspective on you and your medical condition as your long-term pediatric kidney doctor and – more importantly — as a not-so-secret admirer of you.

It is going to be almost 13 years since I met you at Doernbecher Children’s Hospital in Portland, Oregon. You were only 17 months old at the time, a tiny toddler having problems eating and growing. After running many tests and consulting with many specialists, we reached the diagnosis of what was ailing you. You had cystinosis, a “rare genetic disease with no cure.” I remember the surprise, apprehension and love in your parents’ eyes the day we broke the news to them. There were many questions with no sufficient answers about cystinosis. I discussed with your family the challenges facing children with cystinosis from taking multiple medications every few hours, to making sure you received enough calories to grow. And, of course, the frequent visits to our office for follow-ups.

I remember that no matter the changes in your treatment plan I proposed, the response I received from your parents was, “We will do whatever it takes to make Tina better.”

Soon enough you started gaining weight and growing nicely with every clinic visit. I saw a toddler who was full of life, energetic, outgoing and happy. I saw you and your

loving family determined to overcome challenges. I know how hard it had been for you to take so many medications every few hours, especially during the night. And despite all these daily challenges with cystinosis, you were rarely discouraged. Most remarkably to me, you kept embracing life and all the wonders of childhood.

Throughout my professional career, I have been privileged to help care for several children with cystinosis. You stand out among them because you decided to face the condition with vigor and determination. You and your family realized early on that the only way to find a cure for cystinosis was through advancing our knowledge about the disease and supporting scientific research. Therefore, when the opportunity came to participate in the research study for long-acting Procysbi®, you and your family did not hesitate to be a part of it. You helped raise awareness for the condition. Thanks to your partnership with many scientists through the Cystinosis Research Foundation and also through the work of Tina's Hope for a Cure Foundation, we are closer to unraveling the molecular and biochemical mechanisms that cause the disease; therefore we are closer to finding a cure for cystinosis.

THANKS TO YOUR PARTNERSHIP WITH MANY SCIENTISTS THROUGH THE CYSTINOSIS RESEARCH FOUNDATION AND ALSO THROUGH THE WORK OF TINA'S HOPE FOR A CURE FOUNDATION, **WE ARE CLOSER TO UNRAVELING THE MOLECULAR AND BIOCHEMICAL MECHANISMS THAT CAUSE THE DISEASE; THEREFORE WE ARE CLOSER TO FINDING A CURE FOR CYSTINOSIS.**

I look forward to the day when I enter the room for my patient who is newly diagnosed with cystinosis and start my conversation with the family quite differently, "Cystinosis is a rare genetic disease that is curable by the following treatment plan…" That day is closer than we used to think, in large part because of you and people like you.

Tina, it is an honor to greet you and your family all these years and to witness you blossom into a beautiful young lady. Your belief in a better future and your willingness to persevere and look for a cure for your medical condition is an inspiration to all of us.

Thank you for all you have done and will do.

With much love,

Amira Al-Uzri, M.D.

2018 DAY OF HOPE FAMILY CONFERENCE

Because We Dared TO DREAM

by Stephen Jenkins, MD

Day of hope Day of hope Day of hope

THIS YEAR'S DAY OF HOPE FAMILY CONFERENCE WAS AN ENORMOUS SUCCESS. WE HAD 61 FAMILIES FROM ALL OVER THE WORLD INCLUDING AUSTRALIA, SWEDEN, NORWAY, IRELAND AND CANADA. WE WERE EXCITED TO WELCOME 16 NEW FAMILIES, RANGING FROM NEWLY DIAGNOSED INFANTS TO ADULTS.

On Thursday night we got together for introductions. We all shared a little bit about ourselves, as well as our wishes and hopes. Everyone expressed hope for a cure for cystinosis, but also that no matter what happens, that those affected by cystinosis will not feel limited or defined by their disease, and that everybody would lead happy, healthy lives. After introductions, we had delicious Mexican food, something I've come to look forward to every year. Our boys chowed down on chicken quesadillas while their friend, Henry Sturgis, ate his weight in chocolate-covered churros. It was the perfect venue for catching up and getting to know the new families.

MORE

FAMILY CONFERENCE

On Friday morning we got started bright and early. Nancy Stack updated us on the amazing progress of the Cystinosis Research Foundation. Altogether the foundation has raised more than $40 million for cystinosis research since its inception in 2003, funding 164 studies in 12 different countries. It's quite remarkable that such a vibrant research community has sprung up around such a rare disease. After Nancy spoke, it was my privilege to go over the basics of cystinosis at the cellular and organ levels. I have chosen a few of the many outstanding presentations to highlight and share.

Dr. Paul Grimm from Stanford University spoke about the genetics of cystinosis. There are over 100 different mutations identified in cystinosis. The most common is the 57kb mutation, which affects 50-75% of people of Northern European descent. The type of mutations you have determines a lot about how cystinosis manifests. Some people have very severe Fanconi syndrome, while others have very severe corneal disease. Some people don't sweat or tolerate heat very well. If you have the 57kb mutation, you also have a mutation in the receptor that detects heat from hot chili peppers. This gives you the ability to eat very spicy foods.

Dr. Grimm also talked about the upcoming phase 2 trial for a drug called ELX-02 that may allow people with nonsense mutations to make a functional cystinosin protein. It will be administered as an injection, given 2-3 times a week, and may eliminate the need for cysteamine therapy. If you have the nonsense mutation, you should check it out! Dr. Grimm also pointed out that most people in the United States don't get genetic testing because it's usually not covered by insurance. If you would like to get genetic testing done, there are companies like Invitae that charge about $200 to run genetic mutation testing. They will even work with your insurance company to try to get it covered.

Dr. Ranjan Dohil from UC San Diego spoke about the gastrointestinal effects of cystinosis. He showed us his research on how cysteamine itself causes many of the GI symptoms, including a surge of stomach acid production, nausea, and delayed gastric emptying. He also talked about the research that led to delayed-release cysteamine or Procysbi®. He emphasized that fatty foods significantly decrease absorption of Procysbi®, which is one of the reasons it's important to fast before and after. Although the package insert says to fast two hours before and thirty minutes after, he said everyone needs to adjust to their own schedule, lifestyle and needs. Some people may need to wait longer than 30 minutes before eating if they have delayed gastric emptying. Everyone's dose will have to be titrated individually.

Dr. Kathleen Rickert was a

welcome new face this year. She is an orthopedic surgeon from UC San Diego. Although she has not previously seen patients with cystinosis, she has started a collaborative clinic with Dr. Mak at UCSD to see children and adults with bone deformities. She reviewed the common bone complications in cystinosis, including bow-leggedness and knock-knees, and the different medical and surgical ways to manage them. She recommended getting x-rays periodically to monitor the progression of the deformities. I look forward to hearing from her again in the future.

Dr. Julian Midgley from Alberta Children's Hospital in Calgary, Canada, spoke to us about the important and complex process of transitioning from pediatric to adult care. In his unique practice, he is actually allowed to see both children and adults with cystinosis. He recommended starting early and planning ahead; don't wait until the child's 18th birthday. It will be different for each child and each family and should be tailored to the patient's needs.

We heard from Dr. Stéphanie Cherqui on the upcoming phase 1-2 clinical trial for autologous hematopoietic stem cell transplant. She is hoping to submit the IND application to the FDA this summer and recruit the first patients this year. Eligible patients must be over 18 years old and have good organ function. They can have a kidney transplant but must be at least 12 months out from transplantation. Interested candidates will come to San Diego and go through 2-4 days of screening and information. They do not have to commit at that point. If they decide they want to do it, they will come back for 8–9 days for a full, intensive examination, which will include evaluation of kidneys, eyes, lungs, heart, endocrine glands, muscles, bones, neurologic function and quality of life. If they are deemed healthy enough to participate, they will undergo stem cell harvest. Blood is taken and sent to UCLA. At this point, the patient goes back home. At UCLA, the stem cells are modified with a lentivirus vector, which takes about 60-90 days. Once this process is complete, the patient returns to San Diego to get the transplant. This consists of single-agent chemotherapy (busulfan) to make room in the bone marrow. Then the modified stem cells are infused back into the patient.

MORE

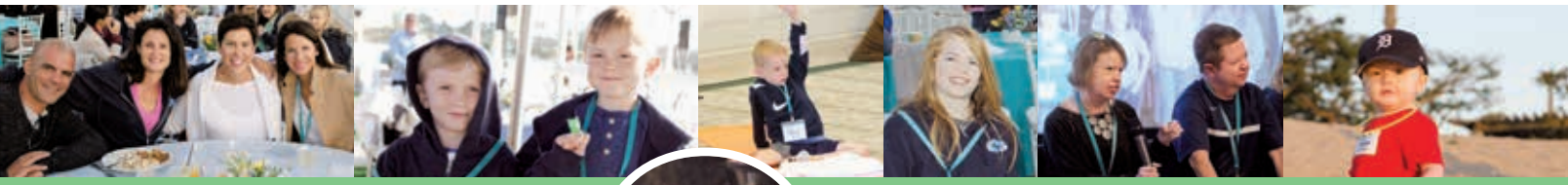
The following month is the riskiest part, as the chemotherapy wipes out your immune system and makes you susceptible to infections. The patient will have to remain in the hospital for one month until the bone marrow recovers. After that, they will remain in San Diego for two additional months with weekly check-ups. Then they can go home but will return every six months for two years to be evaluated. If things go well for the first four patients, then the hope would be to do it in adolescents over 14 years old. I'm very hopeful the trial can start this year. Cross your fingers!

Dr. Bruce Barshop spoke about the new granulocyte cystine test. He showed data demonstrating that overall the newer test has much less variability than the mixed leukocyte test. The goal for the granulocyte cystine test is less than 1.9. The goal for the mixed leukocyte test is less than 1, although that number is apparently a little arbitrary and is based on the upper limit normal for carriers. He unveiled a new shipping kit that will be available soon through Horizon, which will hopefully make the packaging and shipping process much easier. If you'd like a kit contact your Horizon representative.

After a long day of talks, we were ready to get outside and have dinner. CRF arranged for buses to transport us to Back Bay Park and beach, where we enjoyed a fabulous barbeque feast. My kids were super excited for the return of the light up cotton candy wands. After gorging themselves on macaroni and cheese and brisket, they ran down to the beach to start a lightsaber battle. I had to sample every dessert, which included fresh donuts and made-to-order ice cream cookie sandwiches. I think I gained 10 pounds. It was a beautiful setting and a great night to relax.

On Saturday, we heard first from Dr. Mary Leonard from Stanford. She and Dr. Grimm did a study looking at bone health in children and adults with cystinosis. They did a number of tests including strength testing, DEXA, and high resolution peripheral quantitative computed tomography to analyze the muscles and bones. They found that kids with cystinosis have 32% lower leg strength than healthy controls and adults have 35% lower strength. Muscle force was 25% lower in kids with cystinosis, and 13% lower in adults. In people with cystinosis, the cortical bone was thinner and had a lower surface area, and had a 19% lower failure load. The amount and thickness of spongy bone were lower, as well.

Next, Dr. Paul Grimm spoke again – this time about juggling the many aspects of cystinosis care, especially in younger children. He talked about the importance of regular dosing of supplements for Fanconi syndrome,

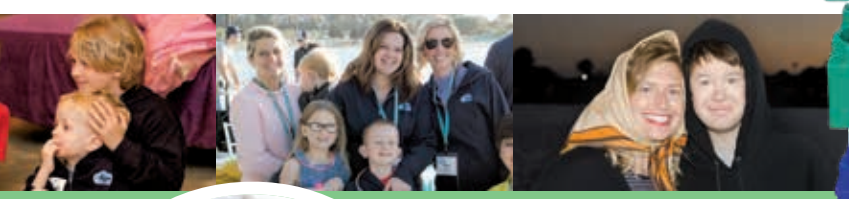


especially phosphorus, potassium and bicarbonate/citrate. He also talked about important medication interactions. Calcium should not be given with phosphorus (neither will get absorbed). Procysbi should not be given with bicarbonate (the high pH will dissolve the beads prematurely). Citrate and bicarbonate should not be combined (it causes a fizzy chemical reaction, which is a good recipe for burping). He talked about indomethacin and using it to reduce fluid and electrolyte losses.

The next talk was from Dr. Doris Trauner from UCSD. I missed her talk, but she reported her results on a study of adults with cystinosis for sleep apnea and its possible connection to memory troubles. The majority of adults tested had sleep apnea, half of which had moderate to severe sleep apnea. This was previously undiagnosed, which suggests that all adults would benefit from being screened. She is still actively recruiting patients to undergo overnight sleep studies.

While Dr. Trauner gave her talk, Dr. Grimm and Dr. Dohil met with the adults with cystinosis, and I met with the kids, ages 11–14. It was very eye-opening to hear these young kids talk openly about their challenges. It was also cool to see how much they support each other. I hope we can have another session next year.

After the last session, we had a question and answer panel with all our researchers and physicians, followed by a question and answer panel with our adults with cystinosis. They discussed how they had overcome many of the challenges of cystinosis and living with a chronic illness. They talked about working, staying active, being compliant with medications, and how to live a full life. Every year I hear from them I am inspired by their courage and hope.



This year's Natalie's Wish Celebration felt like stepping into a dream. On Saturday, April 21, more than 465 guests from across the globe gathered for the cystinosis community's largest fundraiser of the year at the Fashion Island Hotel in Newport Beach, California. The event was a complete success raising more than $3.5 million for cystinosis research in one evening.

As guests entered the ballroom, they were greeted with vibrant shades of pinks, oranges and yellows. Iridescent clouds with inspirational phrases floated along the walls and the colorful table settings created a sunrise radiance across the room. Guests enjoyed a gourmet dinner and decadent desserts, including rainbow cheesecake complete with a unicorn horn and edible glitter! The fantastical atmosphere completely encapsulated the theme: Because We Dared to Dream.

The Natalie's Wish Celebration was the grand finale of the three day family conference, Day of Hope. More than 60 families from all over the world joined the celebration. Guests traveled from Australia, Sweden, Canada, Ireland, Netherlands and Norway to support each other, celebrate the research we have collectively funded, and embrace the strong sense of community we have forged.

The highlights of the evening were the families 'big check' presentations. Fundraising families from around the globe, proudly walked across the stage to present their jumbo-sized checks to Nancy and Jeff. From curling and golf tournaments, to lemonade stands and bake sales, 25 families presented checks totaling $668,000 for cystinosis research. With this support, we will continue to fund the most brilliant researchers and scientists in the world who are committed to finding better treatments and a cure for cystinosis. This community is truly unstoppable!

Over dinner, the room heard from several speakers from the cystinosis community. CRF board member Kevin Partington opened the evening with introductory remarks and a thank you to sponsors and in-kind donors. Nancy and Jeff Stack shared exciting milestones the foundation has reached, as well as provided updates on the scientific research studies the community has funded. At this point, the researchers in attendance were acknowledged with a round of applause for their dedication to a cure and the difference they continue to make in the lives of the children and adults with cystinosis.

Natalie Stack shared a moving update on finishing graduate school and starting her first full-time job. She expressed how it has been difficult




HONORING

the children and adults who are affected by cystinosis and the cystinosis research community for its commitment to our children.


adjusting to the schedule, but how ultimately she is proud to be challenging herself and will not let cystinosis keep her from achieving her dreams.

Nicole Manz, mother of Keegan, shared how he has grown in his journey with cystinosis. She touched on how she admires Keegan's immense courage and bravery, and how he does not let cystinosis slow him down. Though he may have tough days, she expressed that because of CRF she remains inspired and hopeful that we are closer than ever to a cure.

The evening featured a wonderful performance by Eric Hutchinson, who has toured with artists such as Kelly Clarkson, Jason Mraz and Pentatonix. The platinum-selling singer and songwriter played high energy covers such as "Twist and Shout" by the Beatles.

The peak of the evening was the vivacious live auction. Guests bid on a variety of featured items including exciting live events, lavish getaways, exclusive gourmet dinners, and exceptional collections of fine wines. Following the auction, guests had the opportunity to donate directly through Fund-A-Cure. The bid numbers just kept coming. Once the totals were tallied, it was announced that more than $3.5 million was raised for cystinosis. This room broke out in applause and more celebrating ensued. It was a night to remember!



THANK YOU

You have changed

THE COURSE OF CYSTINOSIS

BECAUSE WE DARED TO

dream

2018 NATALIE'S WISH CELEBRATION

The Cystinosis Research Foundation is eternally grateful to all its 2018 Natalie's Wish Celebration donors. With your help we are moving ever-closer to making Natalie's wish a reality — "To have my disease go away forever."

$500,000 AND ABOVE
- Nancy and Jeff Stack

$300,000 AND ABOVE
- David A. Pyle Family

$200,000 AND ABOVE
- Anonymous
- Traci and Tom Gendron

$100,000 AND ABOVE
- Laura Khouri and Michael K. Hayde
- Susan and Thomas F. Moran

$50,000 AND ABOVE
- Christine and William Thormahlen

$25,000 AND ABOVE
- Edelman in honor of Aidan O'Leary
- Jill and John Manly
- Oltmans Construction Co.

$20,000 AND ABOVE
- Gerson and Barbara Bakar Philanthropic Fund
- Dr. Michele and Ed Carpenter
- Lincoln Property Company
- Lugano Diamonds, Idit and Moti Ferder
- Paula and Michael Murphy
- The Redfern Gallery, Beverly and Ray Redfern

$15,000 AND ABOVE
- Anonymous
- Avanath Capital Management
- The Dialynas Family
- Ralph H. Eidem, Jr.
- Bianca and Dave Mossman

$10,000 AND ABOVE
- Joni and Bob Alter
- Nelda Arnold and Dean Henry
- Berkeley Point Capital LLC
- Deborah and Larry Bridges
- Carolyn and C. Preston Butcher
- Contract Carpet Corporation
- Cathy and Bruce Crair
- Donahue Schriber, Bianca and Dave Mossman
- Horizon Pharma
- Betty and Dr. S.L. Huang
- The Allen Hunter Family
- Mary Ann King and Alex Rose
- Janice and John Markley
- Lynne and Augie Nieto
- Sares Regis Group
- Jill and Tom Schriber
- Cindie and Bert Selva
- Silverline Construction Inc.
- TMG Partners
- Linda and Tim Zeller

$5,000 AND ABOVE
- Nancy and Doug Abbey
- Allen Matkins Leck Gamble Mallory & Natsis LLP
- Gerson Bakar & Associates
- Bank of America
- Beall Family Foundation
- Donna and Dac Clark
- D.L.D. Insurance Brokers, Inc.
- Franciscan Friars of California
- Heidi and James Harper
- Susan and Mark Hillgren
- Janet Keller and Bernie Schneider
- Valerie and Bill Kircher
- LBA Realty
- Leadiant Biosciences Inc.
- Karen and Tom Linden
- Lucas Development Corp.
- Barbara and Roy March
- Brenda and Tom Melang, in memory of Sarah Melang
- Tim Moriarty
- Paula and Douglas Neff
- Mark Nimmo, SullivanCurtisMonroe
- Sue Ellen and Paul O'Connor
- C.C. Pascal Charitable Foundation
- Lisa and Chris Payne
- Marie and Anthony Rossi
- Sares Regis Group of Norther California, LLC
- Tejon Ranch Company
- Jill and Larry Tucker
- US BANK

GIFTS IN KIND
- Richard Eddy
- Chef Alan Greeley
- Laura Khouri and Michael K. Hayde
- Idea Hall
- Kokkari Restaurant
- Montage Laguna Beach, Chef Craig Strong
- RD Olson Family
- Pelican Resort, Tom Donovan
- Nancy and Jeff Stack

On Saturday, April 21, 2018, we raised more than

$3.5 million for cystinosis research

$2,500 AND ABOVE
- Cheryl and Dave Ball
- Kay and Michael Bissell
- Bixby Land Company
- Commonwealth Land Title/ Lawyers Title
- Kim and Dick Crawford
- Lisa and Rex DeLong
- Drs. Magdalene and Ranjan Dohil
- Stephanie and Jim Gilly
- Lori and Richard Glucksman
- Beth Anne and Don Haarer
- Brad and Lisa Hillgren, Hillgren Family Foundation Fund
- Suzie and Jim King
- Suzanne and Norman Metcalfe
- Mosio Living Trust, Melinda and Tony Moiso
- Angela and Bill Montgomery
- Natalie and Thomas P. Moran
- Beverly and James Peters
- RL and David Peters
- Susi and Randy Rowe
- Denise and Patrick Russell
- Enza and Jim Saccacio
- Tamara and John Schaefer
- Sandy Sewell, in memory of Sarah Melang
- Chandler Wonderly, Olympus Property

$2,000 AND ABOVE
- Sara Elise-Volbert and Terrance Volbert
- Marcia Gaboury
- The Geraghty Family Fund, Barbara and Edward Geraghty
- Sharon and Terry Hartshorn
- Drs. Joanne and Robert Mak
- Dory and Thomas O'Connor
- Pacific Premier Bank

$1,500 AND ABOVE
- 24 Hours for Hank
- Mary V. Buckingham
- Connie and Ken Coatsworth
- Stephen Lanni
- Camille Longino
- Sherry and Rick Stinehart

$1,250 AND ABOVE
- BJ and Jon Anderson
- Bank of the West
- Lesley and Dr. Alan Beyer
- Mr. and Mrs. D. Robinson Cluck
- Mr. and Mrs. Thomas E. Davin
- Bonnie and Pat Fuscoe
- Gayle and Roy Jones
- Linda and John Kensey
- Robert Lucas
- David E. Martin Architects
- Keiko Sakamoto and Bill Witte
- Cindy and Al Stokke

$1,000 AND ABOVE
- Denice Pace and Dr. Bruce Barshop
- Jean Blum and Carl Lang
- Laurie and Randy Boehme
- Mariette Booth
- Sherry Boucher, in honor of Bailey DeDio
- Karen and Neil Brown
- Jody and Scott Burnham
- Judy and Pastor Dave Christensen
- Diane and Jim Connelly
- Michelle and Reg DelPonte
- Beth and Kevin Farrell
- Feldman Family Trust
- Artyn and Max Gardner
- Chip Harvey
- F. Scott Jackson
- Debbie and Curt Johnson
- Amy and Jeremy Krahe
- Nancy and Ned Kriz
- Sarah Kruer and Jason Jager
- Ethan Matkin
- Barbara and Pat Moore
- Bonnie and Hillar Paju, in memory of Shannon Paju
- Mikel Perskey and Mark Hassman
- Zoe and Don Solsby
- The Stephanie Sorensen Charitable Fund
- The Thomas Living Trust, Cassie and Jim Thomas
- Dr. Doris Trauner and Richard Stanford
- Venture Tax Group

$750 AND ABOVE
- Sandra and Wm. Gary Crouch
- Linda and Kris Elftmann
- Janet and John Griffith
- Anne and John Kittleson
- Gordon McNeill, President Sage Hill School
- Julie and Jack Mitchell
- Suzanne and Stephen Palmason, in honor of the Stack Family
- Lynda and Ken Stack
- Elena and Stephen Stambaugh

$500 AND ABOVE
- Anonymous
- Naomi and Jeff Bailey
- Hinda and Hal Beral
- Marsha and Patrick Brown
- Lora Carpenter and Sean Nicholas
- Stéphanie Cherqui, PhD, and Fabien Faucheux
- Shelah and David Combs
- Cindy and Doug Crocker
- Joan and Jeffrey DeBoer
- Jennifer and James Diaz
- Lara and Peter DiCorpo
- Cyndee and Jon Ely
- Anne and Malcolm Emerson
- Ruthy and David Green
- Donna and Michael Heiken
- Michael Horst
- Pam and Mike Howard
- Aimee and Bob Klaewtanong
- Kathy and Richard Leventhal
- Judith and Robert Lindeman, MD, in honor of Jenna & Patrick Partington
- Nicki and Paul Marx
- Mack Maxwell
- Dr. Anne Marie and Gordon McNeill
- Tom Melang, in memory of Sarah Melang
- Robert Metz
- Marylyn Milburn
- Nancy and Mark Nimmo
- Pam and Steve Quinn
- Zachary Rabosky
- Stephanie and Joe Rentfro
- Angela Rivas
- Jun Sakumoto
- Betty and David Shank
- Shea Properties, Colm Macken
- Ellen Shuman
- Taisha and Zekiele Smethurst
- Justice Sheila Prell Sonenshine and Ygal Sonenshine
- Katie Stack
- Paula Tomei and David Emmes
- Wendy and John Williams
- Julie and Justin Wilson

$250 AND ABOVE
- Anonymous
- Jerrod Blandino and Jeremy Johnson
- Roberta and John Carlile
- Rose and DJ Clarke
- James Inglis
- Ashton and Stephen Jenkins, MD
- Jackie and Kevin Keizer
- Carol and Mark Kelly
- Terry and Phillip LaMotte
- Gina and Vincent Lombardi
- Steve Longo
- Sue and Andy Maguire
- Anna and Charles Mentas
- Larissa Olsen
- Janine Padia and Brian Delahaut
- Ryan Patterson
- Jennifer and Jamie Peachman
- Karen and Frank Ritchie
- Deborah and Frank Rugani
- Janie Tsao
- Joan and Jim Ziegler

$100 AND ABOVE
- Ben Aguilera
- Shaye and Larry Anders
- Lana and Tim Ardaiz
- Julianne and Brad Armstrong
- Nancy and Dennis Bear
- Mr. and Mrs. William D. Campbell
- Corraline and Craig Capelouto
- Sergio Catz, PhD
- Christa and Mike Christ
- Chris Coburn
- Karen and Don Cunningham
- Joan and Robert Curtin
- Kristine David
- Cheri and David DeFillippo
- Christine DeLeon
- Janene and Mark Drilling
- Marthe and Rick Drolet
- Katie Emerine
- Elizabeth Ewart and Dan Roberts
- Richard Fyke
- Lisa Glass
- Lauren Gottlieb
- Christine Grier
- Kimberly Pyke Grimm and Dr. Paul Grimm
- Nicole Groux and Stewie Hartzell
- Richard Groux
- Mary and Jim Harrington
- Cinnamon and Jeffrey Hawks
- Donna and Gerald Johnston
- Jessica and Wayne Jondle
- Erica and Joshua Johnson
- Bethany and Richard Kadish
- Donna Kavanagh
- Lydia Kavanagh
- Molly Keane
- Barbara and Jeff Kulyk
- Melissa and Jim Long
- Ciara and Shane Luke
- Sandy and Jerry Martin
- Dr. Julian Midgley
- Vanessa Milburn
- Connor Mortland
- Anne Marie O'Dowd
- Gail and Marty O'Hea
- Tawnie and Mark Olson, DDS
- Shashona Reedy and Danny Lowery
- Vicki and John Rezzo
- Dr. Kathleen Rickert
- Loretta and Jason Rivas
- Taryn Schneider and Jordan Janz
- Lauren and Robert Silvernail
- Shahla Soltani
- Linda L. Stack
- Paige and Rob Timmons
- Josie and Ed Watson
- Jenny Wichart
- Stuart Winston
- Jennifer Wong
- Dan Wozniak

CYSTINOSIS RESEARCH FOUNDATION | RESEARCH HOPE CURE

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CYSTINOSIS RESEARCH FOUNDATION
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Thank you for your dedication to the global cystinosis community.

The Sixth International Cystinosis Research Symposium

By Stephen Jenkins, MD
CRF BOARD MEMBER,
SAM AND LARS' DAD

I had the privilege of attending the Sixth International Cystinosis Research Symposium held on March 1 and 2, 2018 in Irvine, California. Every two years, all the researchers who have received funding from the Cystinosis Research Foundation get together to share their findings. There were more than 40 scientists from all around the world, including New Zealand, France, Switzerland, Italy, Canada, and the United States. As a Cystinosis Research Foundation board member and someone who has raised money for CRF, it was great for me to see that research dollars are being put to good use. This gathering is important for the researchers, as well, because they get the opportunity to receive critical feedback from other knowledgeable minds in the field, and inevitably the connections they make lead to collaboration.

The keynote speaker was Benjamin Freedman, PhD, from the University of Washington. He is a prominent scientist in the field of induced pluripotent stem (iPS) cells and organoids. His lab has found a way to transform ordinary skin cells into stem cells, which can then be programmed to turn into any kind of cell in the body. His lab turns stem cells into kidney cells, so they can study polycystic kidney disease. These kidney cells can then be manipulated to form something called an organoid, which is like a miniaturized version of an organ. Essentially they grow little kidney-like organs in a dish, so they can study and understand mechanisms of the disease.

Benjamin Freedman, PhD

Alan Davidson, PhD, from Auckland, New Zealand, shared his research in creating induced pluripotent stem cells and kidney organoids with cystinosis cells. These cells and organoids behave just like organs of someone with cystinosis, so they can provide an excellent model to study how different pathways and processes are affected in cystinosis and how they might contribute to the disease manifestations. Dr. Davidson is particularly interested in the mTORC1 pathway, which is important for regulating cell growth and monitoring nutrient availability. It is a pathway that has previously been shown to be dysregulated in cystinosis, and he is looking at different medications that might help. He has also found multiple genes that are upregulated in his cystinosis organoids that are involved in inflammation. This seems to be consistent with the research of others, like Robert Mak, MD, PhD, who have shown the role of inflammation in muscle wasting and other complications of cystinosis. We heard from other researchers who have created other models to study cystinosis, including yeast, fruit flies, zebrafish, and knock-out rats.

There was a whole session dedicated to musculoskeletal disease in cystinosis. Dr. Mak from UCSD shared his research on a hormone called leptin, which regulates energy homeostasis. Too much leptin can lead to energy wasting. Cystinosis mice have high leptin levels, and this may be part of why people with cystinosis have trouble growing and building muscle mass. His lab used a leptin blocker in mice with cystinosis, and the mice had improved bone mass, muscle mass and strength. This could be a promising therapy in the future to treat muscle wasting and bone loss in people with cystinosis.

Anna Taranta, PhD, from Italy, shared her research on the effects of cystinosis on bones. The classical teaching has always been that patients with cystinosis develop rickets, or soft bones, because of Fanconi syndrome, which leads to phosphorus wasting in the urine. Phosphorus is an important building block for bones. She showed that bones in cystinosis mice are abnormal, even before the onset of Fanconi syndrome, and this is likely due to defects in the cells that build and turn over bone. Candice Sheldon, MD, presented the work she has done with Mary Leonard, MD, MSCE, and Paul Grimm, MD, at Stanford. They studied the bones and muscles of 39 patients with cystinosis compared to 182 healthy controls. People with cystinosis had much lower bone mineral density, lower bone strength, and weaker muscles.

Florian Eichler, MD, from Massachusetts General Hospital, shared results from his study on muscle disease in 20 adults with cystinosis. They found that distal myopathy (weakness in muscles of the hands) and dysphagia (trouble swallowing) were very common in adults with cystinosis. Twelve patients reported trouble swallowing and even choking, but only six had evidence of swallowing abnormalities on videofluoroscopy. His study establishes a way to measure outcomes in future clinical trials. Dr. Eichler is also a notable researcher because he is doing a clinical trial using genetically modified autologous stem cell transplant in X-linked adrenoleukodystrophy,

Florian Eichler, MD

a peroxisomal disorder that causes a devastating neurologic deterioration in young boys. This is essentially the same method that Stéphanie Cherqui, PhD, is developing for cystinosis. Dr. Eichler's group has now done the transplant in about 30 boys and has seen remarkable results, with most boys experiencing a halt in the progression of their disease. Talking with him made me very excited for the upcoming stem cell transplant trial for cystinosis!

Pierre Courtoy, MD, PhD, from Belgium, shared his research on a protein called megalin in cystinosis. Megalin is a protein that allows the proximal tubule cells of the kidneys to reabsorb proteins. These proteins have a lot of the amino acid cysteine forming disulfide bonds in them, so if they get reabsorbed and metabolized by kidney cells, this can lead to lots of cystine deposition in the kidneys, which causes kidney disease. When he and his lab knocked out this protein in a mouse with cystinosis, they saw cystine levels go down, with no cystine deposition in the kidneys. This led him to hypothesize that inhibition of megalin could be a treatment for cystinosis. It turns out that supplementation with arginine and lysine inhibits megalin. They are studying whether feeding mice arginine and lysine supplements helps prevent kidney damage in cystinosis. This could be another promising therapy.

Francesco Emma, MD, from Italy shared updates on his research using drug library screening. His lab had previously screened more than 1,200 drugs to see whether they had an effect on cystinosis cells. They found one promising prospect, disulfiram, which lowered cystine levels like cysteamine. This was exciting because disulfiram has been around for a while and is super inexpensive. Also called Antabuse, it's a drug that makes people sick if they drink alcohol, so it's used to treat alcoholism. He tested it in cystinosis mice, and unfortunately it made them very sick, with poor growth, hair loss, and other toxicities. They tried reducing the dose, but at lower doses the drug failed to reduce cystine accumulation. They then tested the drug in zebrafish with cystinosis, and it caused significant birth defects. What started as a promising new therapy became a cautionary tale of why it's so important to do research before trying new medications on humans. Fortunately, they have two other compounds, which are available over the counter, that have shown to be safe and beneficial in cystinosis mice and may treat Fanconi syndrome. He needs to do additional studies before they can be tested on humans.

Paul Goodyer, MD, from Montreal, is working with a company called Eloxx on a clinical trial for treatment specific to cystinosis patients who have a nonsense mutation. A nonsense mutation is when there is a single base pair change in the DNA that leads to a premature termination of protein synthesis. It's like a big STOP sign. Eloxx has developed an injectable drug that allows the cellular machinery to ignore this stop sign and continue synthesizing the cystinosin protein. This is a very intriguing idea for therapy and could provide a very effective treatment for the 20 percent of the cystinosis population that has a nonsense mutation.

Vincent Stanton, MD, from Thiogenesis Therapeutics, shared his exciting research on creating a better form of cysteamine. They modified cysteamine by adding a molecule to form a new compound, a prodrug or precursor to cysteamine. This compound is metabolized in the gut to produce cysteamine over at least six hours. Cysteamine is absorbed as it is created, with some produced in the first part of the intestine, and more metabolized and absorbed further downstream. This results in a much lower peak concentration (which is expected to reduce the side effects of odor and nausea) and a more sustained blood level of cysteamine (which should extend the time between doses). They found that a large reservoir also ended up in the colon of rats, where it continued to be absorbed. This may result in a medication that could be taken once or twice a day, with better efficacy and fewer side effects than Cystagon® or Procysbi®.

Dr. Cherqui presented an update on her upcoming clinical trial for the autologous stem cell transplant. She is hoping to submit the IND in the fall. The first trial will include six patients. We are hopeful the trial will start later this year!

Morgan Fedorchak, PhD, from the University of Pittsburgh, presented her research on a controlled release cysteamine eye drop. She has developed a thermo-responsive hydrogel that turns from a liquid to a solid when it touches the eye. It is filled with small microspheres that are loaded with drug. She has made a similar system for glaucoma drugs that can be taken once a month. She has found that cysteamine is a much more challenging drug to work with, but is hopeful that the drops could be done once a day. She is currently studying the drops in rabbits and will soon start testing it on cystinosis mice.

Sihoun Hahn, MD, PhD, from Seattle Children's Hospital, spoke about his work to develop a newborn screening test for cystinosis. They have found a way to detect the very small amounts of cystinosin peptides in the blood. If no cystinosin peptides are detected in the blood, then it would be considered a positive screening test and would lead to a white blood cell cystine test. His next step is to validate the study in about 100 cystinosis patients, and then they

RESEARCHERS (L-R):

Pierre Courtoy, MD, PhD • Daniel G. Bichet, MD, MSc • Francesco Emma, MD • Julie R. Ingelfinger, MD
Corrine Antignac, MD, PhD • Stéphanie Cherqui, PhD • Sergio Catz, PhD

will do a large scale pilot study, probably in the state of Washington. Early detection can make a huge difference in cystinosis, since starting cysteamine therapy as soon as possible can delay and even prevent many of the complications of cystinosis. I also spoke with Dr. Hahn personally about the so-called "mitochondrial cocktail." As a pediatric geneticist, he treats many children with rare mitochondrial disorders, for which there are no good treatments. For these children he tries vitamins and supplements which are necessary for mitochondrial function, including Coenzyme Q-10, vitamin B complex, vitamin E, and levocarnitine. He said that there is clearly mitochondrial dysfunction in cystinosis cells and that it wouldn't cause harm to try these supplements to try to improve mitochondrial function, and therefore hopefully improve neurologic and musculoskeletal health. He said if he had a child with cystinosis, he would definitely try these supplements. He did mention that Coenzyme Q-10 comes in two forms: ubiquinone and ubiquinol. Ubiquinone is cheaper and the most popular version found in stores. However, it is not absorbed well and is felt to be less effective. Ubiquinol, on the other hand, is absorbed much better, but is more expensive. He recommended using Ubiquinol.

Sergio Catz, PhD, from the Scripps Research Institute, presented his research on how cystinosis affects chaperone mediated autophagy. Autophagy is an essential process through which cells recycle damaged proteins and organelles to generate nutrients during periods of starvation or stress. Dr. Catz has shown that even with cysteamine treatment, autophagy remains impaired. They are working with another scientist, Ana Maria Cuervo at Albert Einstein College of Medicine, who has discovered a compound called CA77, which stimulates autophagy. Dr. Catz has tested this compound in cystinosis cells, and they have improved survival during periods of stress. He is going to test this compound in cystinosis mice, with the help of Dr. Robert Mak's lab.

Robert Mak, MD, PhD

There were many other talks and posters, and I wish I could include them all. It was incredible to see so much research in action. It was exciting to see how studying the basic science of cystinosis has led to many new potential therapies. I was filled with hope that my two sons will be able to live long and healthy lives thanks to the tireless work of this scientific community and the dedication of the Cystinosis Research Foundation.

Turning my life around

By Samantha Grover
EXETER, NEW HAMPSHIRE

When something bad happens to you, one of three things happens: it defines you, destroys you or it strengthens you. My journey has traveled through all three phases. When I was 18 months old, they discovered I had rickets. This led to a diagnosis of Infantile Nephropathic Cystinosis. It defined my life.

My childhood was different from my peers and my siblings. My parents were super over-protective. I was not allowed to go out and do things like other kids, and I always had to go to bed very early. I couldn't go to sleepovers because I wet the bed. I would have to get up in the middle of the night starting at age 8 or 9 to change my bed and wash my sheets. I was finally able to go to a sleepover at a close friend's house when I was 11. But I had to wear a diaper. My friends were wearing "real" bras and I was still in diapers! Then there's the Cystagon®. I hated everything about it. I hated the taste and the smell. I still can't handle the smell or taste of grape juice today. Cystagon® would get stuck in my throat and start to melt, so I'd throw up and have to take it all over again. I was embarrassed by constant trips to the nurse's office to take my eye drops and hurrying to class so I wouldn't be late and have everyone stare at me as I walked in. When I was in sixth grade I was smaller than my peers. I was a cheerleader and had to wear a skirt made for elementary school kids. A teacher held up my skirt for the whole class to see and pointed out that it was not like the other girls' skirts. I was so adorable and cute. It was super embarrassing. I would hear other kids talk about how I smelled bad and how my breath smelled bad and I should start brushing my teeth. But I was brushing my teeth after every dose of Cystagon®. The good thing is I have fantastic teeth and have never had a cavity, but hated being different.

When I was 12, my mother donated a kidney to me.

I HAD FOUND "MY PEOPLE!" A COMMUNITY I COULD BELONG TO.

Finally, no more bed wetting! I was finally going to be just like everyone else, or so I thought. But I still had eye drops, rejection medicine and Cystagon®. I was like other kids in some ways though. I constantly fought with my step-mother and my dad had to force me to take my medicine. Throughout high school I was still very sheltered. After all, I wasn't supposed to live this long. I knew what medications I was taking, but I never really understood what they were for. I knew I had cystinosis, but I thought it was a kidney disease. I even went to Camp Sunshine on renal week with my family. My kidney was my only problem.

When I graduated from high school, I was excited to move out of the protective and oppressive shelter of my childhood home and become an adult. I moved to North Carolina to be near my mother and began my life my way. I got an apartment, a job and started to go to school. I even got my first boyfriend ever! As an adult, I was able to make my own decisions, and I decided I didn't need to take my medications all the time. After all, it was just a kidney disease and I was still taking my rejection medicine. When you've stared death in the face several times before you turn 18, you get the "I am invincible" complex. I'd get sick but I'd always get better. My life was wonderful. I went out with my boyfriend and my friends. I went to bed whenever I wanted and life was wonderful. Then my mother decided to move to Florida and she decided that I was going with her. I loved my life, my friends and my boyfriend and I was not going, no matter what. So, while I was not home, she packed up my apartment so I would be forced to go. I was an adult. I was going to make my own decisions. I was staying. So, I moved in with friends and off she went to Florida with my things. She thought I would be forced to follow her. I'd show her though, not only would I stay, I would stop taking my rejection medication and get

ELLEN DEGENERES INSPIRES ME TO DO BETTER, TRY HARDER, HAVE A LITTLE FUN AND IS A HERO TO ME. BEST WAY I KNOW TO STOP CRYING IS TO WATCH ELLEN.

this stupid kidney that was hers out of my body. After all, I had five other matching family members and I didn't need her kidney. I was an adult and it was my adult decision.

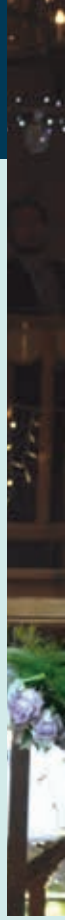
Cystinosis was now destroying my life. The hospital in North Carolina called my dad and let them know my kidneys were failing. Without hesitation, he immediately drove straight down and brought me back to New Hampshire. I did not want to move back home where I would be treated like a little girl again. So, I decided to move in with my grandmother, my aunt, and my cousins; they would treat me like an adult. I began dialysis on January 31, 2008. I did not like dialysis. It was all elderly people and I was only 19 years old. This was only the beginning of my descent; my adult decisions would continue to destroy me.

My life was a rollercoaster. I had great days, not so great days, and hospital stays. I moved in with friends and tried to pretend I was just like everyone else, only I had dialysis three days a week. Eventually I moved in with my aunt and tried to get my life back on track. I even tried to get on Procysbi®, but due to muscle wasting in my throat I was only able to swallow two pills with a bottle and a half of water. Not acceptable for someone on dialysis. That was almost my entire allotment of liquids for the day. I was feeling pretty good otherwise, so I decided not to worry about it. My uncle and aunt helped me to get an apartment with a friend and life was looking great again. Perhaps my rollercoaster ride was over.

My lease was up, my friend was moving in with her boyfriend, and my aunt had remodeled me out of a bedroom, so back home I went. My parents had also remodeled their house, so they had their own new apartment above the garage. Unfortunately, this also meant I was moving into the three-bedroom house with my newly married older brother and one of my younger brothers. The only thing that made it bearable was I was sporting a brand-new bedroom set. I hung out with my aunt and cousins when I wasn't at dialysis because all my friends had jobs, boyfriends or were married with kids. I only went back home to sleep. I was an adult and hated being back home.

Depression set in. I hated my living situation, going to dialysis, lack of friends, and was just drifting through every day. I felt like I had lost almost my entire support system because I was "an adult." Ellen DeGeneres became one of the biggest members of my support system and she didn't even know it. She inspires me to do better, try harder, have a little fun and is a hero to me. The best way I know how to stop crying is to watch Ellen. Then the other members of my support system swooped in once again to turn things around. They got me a safer vehicle for New Hampshire weather, an apartment right next to my cousin and furnished the entire thing. My depression began to lift once again.

I had never heard of a Patient Access Manager (PAM) before, but it turns out I had one. We discussed my inability to take Procysbi®, and we discussed a feeding tube just for medication. I was excited that I might be able to get back on track again. She also told me about a town hall meeting in New Jersey where I would meet others with cystinosis and their families. I had never met anyone with cystinosis before. I was so excited but my social anxiety had me petrified at the same time. I was very happy that my aunt and uncle



agreed to go with me. I would need their support to be able to survive the weekend, even though my PAM would be there as well.

As luck would have it, dialysis gave me my flu shot the day we left for the town hall meeting. As if social anxiety was not enough, now I had a slight case of the flu, complete with fever, body aches and nausea. My aunt knows me very well and not once the entire weekend did she allow me to use the flu (fear) as an excuse to not attend a session. I am so grateful that she didn't. I was so thrilled to learn that I was not alone. There was a way for me to relate to each person in one way or another. There were people that were bullied, wet the bed, smelled, were noncompliant or embarrassed just like me! I had found "my people!" A community I could belong to. I was not feeling so alone and different anymore. They were friendly and made me feel so welcome. I learned about cystinosis; it's not just a kidney disease. I wasn't the "sick kid" anymore, I wasn't sick and it wasn't my fault. That was very eye-opening. Best of all, I learned about Day of Hope and Natalie's Wish and I was determined to go. My only regret was that the flu shot made me slightly out of it and I wasn't able to participate more.

Day of Hope was everything I could have hoped for and more. My aunt, uncle and cousin attended with me and not only did we learn so much about cystinosis and the research that is being done, we had fun. We met so many friendly people, made friends from other countries and truly embraced the feeling of support that such a community bestows. The Natalie's Wish event was wonderful! I was brought to tears by the stories that were shared and the generosity of those that attended. I now felt empowered to be an advocate for myself and my medical care. I was ready to go home, get my medical team on board, and charge forward. Day of Hope was altering my life in such a positive way.

I came home invigorated with concrete ideas to improve my quality of life. But once again I'd somehow stepped into a rollercoaster. What I had thought was a buildup of fluid that dialysis was not removing turned out to be ascites. The fluid not only made it difficult to breathe from the pressure on my diaphragm, it also made me unable to eat very much as it was also pressing on my stomach. I dropped a lot of weight and began to look like a skeleton who was eight months pregnant. My hair was also falling out and it was so thin, you could see my scalp. When I got a new kidney, I didn't mind the scar, it was why I was alive. My fistula was for dialysis; it keeps me alive. My feeding tube allows me to take my medication, and I can hide that. But I couldn't hide my hair or my frail body. I now looked like the "sick kid." I got a wig to cover my head and everyone thought I was a cancer patient. I got so weak I was forced to stop driving most days and had to rely on others to drive me to and from dialysis. Not an easy task when your support system is so small. I hate to ask for help so often my refrigerator was empty because I hated to ask someone to take me shopping. While I had rides to get to my doctors' appointments in Boston, when my aunt was away it was sometimes difficult to get someone to go with me and I had to go alone. My quality of life was not good.

Day of Hope

WAS ALTERING MY LIFE IN SUCH A POSITIVE WAY.

I CAME HOME INVIGORATED WITH CONCRETE

IDEAS TO IMPROVE MY QUALITY OF LIFE.

My words and actions were contradicting. I said I wanted to get better, but I was cancelling doctors' appointments, still not taking meds and feeling very sorry for myself. I was denied a kidney and liver transplant because I was noncompliant. I felt like no one ever visited me and no one wanted to go anywhere with me. My aunt was traveling a lot and I was so lonely. My mother came to stay with me to try and get me on track with my father's help, but that ended up being a total disaster. One of my friends was torn between being a friend, my housekeeper and a mother figure. My sister used to come over and hang out with me a lot, but now she was mainly coming over only to drive me to and from dialysis and walk my dog. My brother moved in with me to help me out, but all I focused on was that he was a slob and he was sleeping in my craft/meditation room. My sister-in-law tried to organize a girls-only party but I only focused on the fact that everyone only stayed for a couple hours and then went home and complained no

one ever came to visit. Two of my aunts and my sister had to sit down and let me know that no one wanted to stay because I was rude and nasty to everyone. Nothing was ever good enough for me. My attitude was driving everyone away. I was blaming everyone else for my circumstances and was finding fault in anything and everything. My words meant nothing because my actions were the complete opposite.

I had now hit the lowest point in my life, which is interesting considering how many times I had been close to death. Through my depression and anger with my life and situation, I had alienated my support system and those who loved me most. I'd driven away the people who have always been there for me and was now challenged to get them to return to my life. I missed them so much. I was dying because I was too depressed and too afraid to live! What could motivate me to crawl from the lowest point in my life and scale this mountain of depression, anxiety and fear?

When I was 2 years old, one of my aunts died in a tragic accident. Unfortunately, I don't remember her. But I do know her through the stories and memories of others and pictures they have shared with me. My beloved niece is 18 months old. I don't want her to remember me through other people's memories, I want her to remember me because she is old enough to create memories of us together. It is now time to seriously fight for my life, prove the doubters wrong and get back on track. I want my niece to remember me through her memories and learn to enjoy my love of food, music, crafting and of course, Ellen.

Cystinosis has become my strength. It has made me who I am today. I believe every challenge I have gone through has made me not only a stronger person, but a better person. I now appreciate life so much more. I want to live. I am still afraid of things other people take for granted. I am a hard stick and my veins are shot. If I need medication right away or IV fluids, they have to bring in the ultrasound in order to find a vein. That typically takes 30 minutes or more, not a good thing if my life is on the line. But I am determined to live, to be compliant. I will look ahead to the future and not worry about yesterdays. I will live each moment to the utmost. I will make my friends and family proud of the battles I have faced and remind them how much their support has meant to me, especially when it was hardest to stand beside me despite my words and actions. I don't want to disappoint them yet again, I want to be remembered for the good things I have done, how much fun I am to be around and how I made a difference in the world. I want to give back to the world through mentoring, volunteering and spreading awareness of cystinosis. While each day remains a challenge, I will face it head on with open arms. Life's short for everyone, if you're not enjoying every moment while you are alive, you are pretty much dead.

AIDAN'S JOURNEY BEGINS

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

February 28, 2016 was the best day of our lives; it was the day we welcomed our beautiful baby boy, Aidan James O'Leary, into this world. Aidan seemed healthy, vibrant and energetic right from the beginning. He continued to thrive until about 8 months, when we noticed some drastic changes in his eating habits, growth, and energy level. After countless trips to the pediatrician, an emergency room visit, and a grueling month-long stay at Lurie's Children's Hospital, our little Aidan was diagnosed with nephropathic cystinosis at 11½ months old.

The doctors warned us that this disease often requires more medication than nearly all other known diseases, and they were not exaggerating. Aidan takes 18 medications every day. His appetite is variable – mainly as a side effect of his medications – therefore, we also feed him a blended diet using his G-tube to ensure he gets proper nutrition and continues to grow.

As all cystinosis families know, living with this condition is a challenging journey. It's been one year since Aidan's diagnosis, and there have been many triumphs and tribulations. The majority of our last year was centered around gathering knowledge, endless advocating, building the best medical team, titrating medications, managing side effects, washing syringes, making blends, and trying to figure out our "new normal" as a family. Jim has been amazing with balancing his executive responsibilities and increased demands at home. I quit my job as a physical therapist and have assumed the role of a stay-at-home mom/nurse managing Aidan's care. I spend most of my days chasing around a very active 2-year-old who has figured out that his medications make his "tummy hurt." Needless to say, the art of distraction has become my best friend.

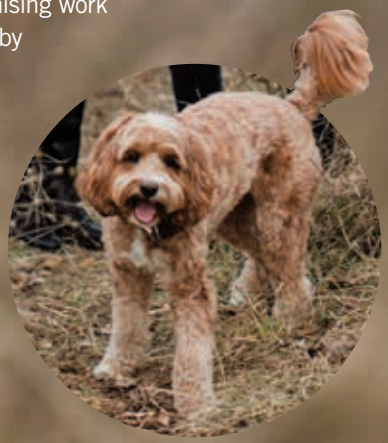
We have just now started to emerge from our "PTSD fog" following diagnosis: learning how to live with cystinosis versus letting it completely dictate our path. We hope to heal. As many of you know the road to/through diagnosis is an emotional and physical rollercoaster that can leave deep scars. We're trying to make peace with the medical missteps and suffering Aidan endured. It amazes us that despite everything Aidan has been through, he wakes up every day with a huge smile on his face, happy to conquer another day. He especially loves music class, his dog Gus, and causing just enough trouble to keep us on our toes. Little Aidan's nearly permanent smile is infectious, and we're thankful that he's doing so well. However, we realize that our journey with this disease has just begun.

We're thankful that Aidan was diagnosed before his first birthday – which is relatively early – and still has most of his kidney function. We are ecstatic to report that with his rigorous medication, feeding, and electrolyte replacement regimen – Aidan is not only stable but thriving!

As we unfold this new chapter of our lives, we have found new purpose and drive – helping to find a cure for cystinosis. The O'Learys and our amazing friends have begun raising funds. We have raised over $40,000 to date, and our first charity golf event, "Aidan's Army Golfs Fore a Cure," is scheduled for August 27 at Forest Lake Country Club in Bloomfield Hills, Michigan.

We're forever thankful to our family, friends, and the broader cystinosis community for supporting us this past year. Thank you to the Stack family for your unwavering leadership.

We're hopeful for the future, mainly due to the outstanding and very promising work being funded by the Cystinosis Research Foundation. Thanks to your efforts, a brighter future is imminent for our children.



AIDAN'S DOG, GUS

IT AMAZES US THAT DESPITE EVERYTHING AIDAN HAS BEEN THROUGH, HE WAKES UP EVERY DAY WITH A HUGE SMILE ON HIS FACE, HAPPY TO CONQUER ANOTHER DAY.

An Ode for the Cause

By Denice Flerchinger, Tina's mom
CLARKSTON, IDAHO

Our beloved mother, Barbara, passed away on Christmas Day. Mom was a true pillar of faith, and she shared that faith with all her children and grandchildren.

Our youngest daughter, Tina, would spend many summer days and school breaks hanging out with her "Grandma Barb." They had a beautiful connection. They could be found praying the rosary, reading, gardening, watching "The Price Is Right," or napping together. Grandma taught Tina how to play pinochle and both were equally competitive.

Mom enjoyed reading *Cystinosis Magazine* and always kept up-to-date on the latest happenings in the cystinosis community. She looked forward to attending our Tina's Hope for a Cure fundraising events and was a generous giver to the cause. To continue mom's giving spirit, family and friends donated over $5,300 to Tina's Hope for a Cure in her memory. Mom would be so pleased.

Mom always told Tina to have faith and trust that God would cure her and her cystinosis friends one day. We have no doubt our beautiful mother is watching over Tina and pleading with God for that cure that we have hoped and prayed for.

gel

GRANDMA BARB AND TINA HAD A BEAUTIFUL CONNECTION


2400 FEET OF
SCHWEITZER
LODGE TO LODGE GIANT SLALOM
WE COME TOGETHER, RAIN, SNOW, OR SHINE... WE CELEBRATE AND WE COMMIT TO FINDING A CURE!
HANKS
HANKS
HANKS

24 HOURS FOR HANK TURNS NEAR-BLIZZARD CONDITIONS INTO A FLURRY OF DONATIONS

By Kristin Soures,
Board Member, 24 Hours for Hank
SANDPOINT, IDAHO

We just completed our 10th annual 24 Hours for Hank event at Schweitzer Mountain Resort. On March 24, 2018, travelers from near and far came to help us celebrate this amazing day. This year was exciting and sentimental for many reasons. We were able to reflect on the last 10 years and celebrate with our participants some pretty epic memories!

This year we changed things up a bit. We aren't sure if it was because we are getting too old for 24 hours of non-stop skiing, or if we just decided to try something new. Whatever the reason, it was an astounding success! This year, our participants got to partake in a truly memorable experience. They got to ski 2,400 vertical feet down 2.4 miles in the longest giant slalom course in the United States! And, they did this in near blizzard conditions.

The race started at sunrise, two hours before the resort is even scheduled to open. Once again, we were challenged with weather, and though all of the snow was not ideal for racing giant slalom, it made for some epic powder skiing the rest of the day. The weather never detours our participants, and they come anyway. They come because we are all committed to finding a cure. We have raised over $1.2 million to cure cystinosis. This year we set a goal to raise $150,000. We not only achieved this goal, we exceeded it!

This accomplishment is because of the amazing people who come to support this worthwhile and important cause. Some came for the first time ever this year and we welcomed them. Others have been with us over the years, and we were able to honor those that have been with us all 10 years. We spent the day together starting at sunrise and skiing down this amazing course, eating breakfast together at the Outback lodge (another new first), participating in some pretty intense challenges, skiing in some epic powder, honoring our winners with awards, eating some amazing food, drinking some pretty great drinks, bidding on fabulous auction items, honoring Hank and all others affected by cystinosis with an emotional video, and dancing the night away at the after party.

So many of our participants have come to be like family to us. They travel from all over the country and from their near backyards to be with us and support us in finding a cure. Schweitzer Mountain and its people have been so good to us, and they have allowed us to create this very special bond. It truly is a magical time. We come together, rain, snow, or shine. We ski hard, laugh hard, play hard, and even shed tears. We celebrate and we commit to finding a cure! Thanks everyone for making this year yet another success!

A DAY IN THE LIFE
WITH CYSTINOSIS

On Friday, February 2, I was so moved by events of the day that I jotted down some notes, thinking I might soon expound upon them in my update for *Cystinosis Magazine*.

It was a day that saw our family in the throes of sickness and recovery. Jenna was sick. I was sick. Patrick had his second knee surgery at Shriners eight days prior, and was clumsily (and very bravely) moving about in a wheelchair. It was a day of contrasts: frustration and kindness, of sickness and sweet inspiration. All of these emotions somehow related to our family's collective journey with cystinosis:

JENNA

We ultimately learned Jenna had mononucleosis. Diagnosis took two weeks. It is common, but not as cut-and-dried when you combine it with cystinosis. My car was on autopilot to the doctor's office. Blood work was done over and over. She was needy. Caring for Jenna felt like caring for a newborn.

PATRICK

Patrick and I had set a goal to "get to school on time today." Alas, we were one minute late for the bell, underestimating the time it takes to load in and out of the car with a wheelchair. Patrick's friend Matthew saw us at the school drop off and helped wheel his buddy into school. Patrick hauled his overstuffed backpack and a big box of Pampers for donating to the school diaper drive in his lap. The principal, Mrs. Faires, was offloading kids that day, patiently assisting us in our efforts with Patrick. He and Matthew would not be counted tardy. She told me that the whole school was praying for Jenna.

A TENDER SURPRISE

By 9 a.m., we are running late for Jenna's doctor appointment. She is exhausted, can hardly get out of bed, and she has a 103-degree fever. I have the flu, and it's making me frustrated, tired and grumpy. My phone rings, and I accept a call from an unknown number on hands-free in my car. It is Jenna and Patrick's kindergarten teacher, Dona Pollacchi, calling to ask me a question. "Do you have a minute, Teresa?" She has called me only a few hours after her husband has passed away. She wonders: "Would it be alright for me to list Jenna & Patrick's Foundation of Hope as the charity to give to in Leonard's memory?" I cry in the car out of love and heartache for Dona. I am moved by the mark my kids made on Leonard's heart, that he would make this request before he died. I am given a big dose of perspective. I am grateful and my kids are so, so strong. We get through Jenna's appointment. She is severely dehydrated and needs IV fluids as soon as the infusion center can accommodate her. Kevin makes changes to his work schedule so he can be home with Jenna and I can go to my own doctor appointment. I need Tamiflu, and I need to pull it together!

BUOYED BY COMMUNITY

We are preoccupied with Jenna, who is so sick she commands our constant presence and attention. Even at night, Kevin and I play musical beds and take turns sleeping with Jenna beside us. Patrick should have been walking about three days after surgery. Nine days later, and there is no sign of independent walking for Patrick. He has never used the muscles required for walking in this "new way." He is learning to walk again, for the third time in his life. Every movement and task is an effort for all of our family (the bathroom, the stairs, the car). Kevin and I wonder: Is Patrick doing OK? Is his temporary disability hampering his school work and ability to participate and enjoy his friendships? In early afternoon, I receive a call from the school. It's Patrick: "Mom, my friends wonder if they can push me in my wheelchair to Vic's Ice Cream Parlor after school?" My heart leaps. "Of course, Patrick," I tell him. "Have fun!" He is unabashedly wheeled by a troupe of boys and girls who fight to push him. I know this, because parents from our neighborhood saw this, were heartened by it, and felt compelled to call me. It's as if they knew my worries. We are constantly buoyed by the loving and supportive community that surrounds us.

A VIVID REMINDER

Jenna has missed 21 consecutive days of school. Some days she only got out of bed to use the bathroom. Her breath was metallic, her lips split from dehydration, she lost 18 pounds. Jenna had four blood draws and four days of IV infusions, which were necessary to keep her hydrated, and to protect her already damaged kidneys. She charmed her pediatric infusion center nurse, in spite of feeling miserable. On all four visits Jenna was cared for by "Nurse Melinda" at UC Davis, who claimed Jenna as her patient the minute she showed up on the schedule. Sitting beside Jenna as she was hooked to an IV, resting in a hospital bed, proved a vivid reminder of those days and months 12 years ago, when cystinosis was a word we'd never heard.

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

THE PRESENT

As I write this, it is February 21. Patrick is still using a wheelchair but he is making great progress, thanks to the rehabilitation services at Shriners Hospital in Sacramento and thanks to Patrick's unrelenting efforts. He is a patient and determined kid, processing all that he deals with quietly, privately. Patrick is surely aware of his unique, difficult, beautiful lot in life. He is profound and spiritual, a dear friend to many. And he bickers with his sister constantly!

Jenna has bounced back! Did I ever question that she would? She is back at school, eating heartily and making jabs at her brother whenever she can. Jenna focuses on all that is fun. She likes to do what makes her happy and to see others happy. She finds the nightly news and many books, movies and TV shows "too sad." Jenna has faced enough in her days that we feel little need to force the weight of the world upon her. Most of her strength is used for keeping herself healthy and seeking happiness – encouraging anyone around her to share in day-to-day, simple joys. An episode of "I Love Lucy" while having her arm tickled is about as good as it gets for Jenna. Such contentedness is a gift...until it's time to do homework, which is not joyful.

STRANGER THINGS

The Netflix series that has seen us through all of this. Each episode found us calm and riveted by something other than knee surgery, mono, work, flu, school and worry.

"Stranger Things" is scary! But, not as scary as February 2018!

Love and endless thanks to our family and friends,

Teresa, Kevin, Patrick and Jenna Partington

PATRICK IS A PATIENT AND DETERMINED KID... HE IS PROFOUND AND SPIRITUAL, A DEAR FRIEND TO MANY.

JENNA USES HER STRENGTH TO SEEK HAPPINESS – ENCOURAGING ANYONE AROUND HER TO SHARE IN DAY-TO-DAY, SIMPLE JOYS.

CYSTINOSIS NEVER TAKES A VACATION. IT IS AROUND-THE-CLOCK, EVERY SIX HOURS (WE ARE STILL ON CYSTAGON®), DAY IN, DAY OUT, THROUGH ALL FOUR SEASONS.

Left to Right: Madelyn (4, with cystinosis), Sara (10), Aliyah (7, with cystinosis)

Taking Cystinosis On Vacation

By Crystal Walker, Sara, Aliyah and Madelyn's mom
CALGARY, ALBERTA, CANADA

The countdown is on. Three days until a warm Mexican vacation with our entire family, away from the snowy tundra that is Calgary, Canada in January. The suitcases have been packed, unpacked and repacked countless times as we debate over sweaters and rain-jackets, (who's kidding, anything over 50 degrees Fahrenheit is bikini weather right now), check off our list of travel medications, (Tums, anyone?), and ensure that we have enough bathing suits for five people for seven days of swimming.

In addition to all of the regular questions of regular travelers, we have an abundance of other concerns. Really, it doesn't matter if we forget anything that we can buy in Mexico, but cystinosis meds? We have to be 100 percent certain that we have exactly what we need to keep our kids healthy for the next seven days, and exactly what we need to cross two borders with way more liquids than travelers are allowed!

We have a large thermal tote packed with liquid formula and five bottles of different electrolytes for our two younger daughters Aliyah, 7, and Madelyn, 4, both of whom have cystinosis. Three sets of bolus syringes and G-tubes in case we lose one or it gets clogged. Multiple Ziploc bags to keep dirty syringes, and a small thermal bag for daytrip meds. Wipes to clean off the hose and a bottle brush.

So now the big questions begin:

- Do we prep medical syringes for seven days or only five? (How often do we want to be washing syringes and prepping meds?)
- How many Cystagon pills do we need for the two of them for seven days, and how many extra days' worth do we bring?
- Do we need Pedialyte in case someone is throwing up?
- Do we have accurate letters depicting their condition and meds for border guards?
- Who is going to carry that heavy liquid tote?

And there are logistics to consider. Do we adjust the dosing schedule for our six-hour flights? We are on vacation, and both Bob and I would like to sleep through the night – so what's the best way to ensure we rest? We decide to alternate nights, which works out well. We make sure we have our chargers since I don't trust anyone else with my alarm, and anyway, the loud ring of a hotel phone is likely to give us all heart attacks at 2 a.m. Finally, we are sure we have everything (we find out later we forgot the bottle brush). True story, though, my heart palpitates when we leave the house as I check my mental checklist – again – that we have everything we need for a healthy trip. Thoughts of hospitals and emergency procedures are never far from my mind.

Cystinosis doesn't take a vacation, even when we do. We bring multiple doses of meds to the beach and back again, and to each excursion, including emergency G-tube kits in case one pulls out while drifting down an underground river. Occasionally, Aliyah sits back in a lounge chair with some nausea while her cousins splash in the pool. One night, there is a lot of vomit and we are grateful for tile floors! An entire counter is taken up by medical supplies and we wonder if we have to tip differently when we request a LOT of extra water! Without a freezer in our room, we make the trek to the front desk several times for cold ice packs. Our extended family kindly waits for us when we prep and administer meds in the morning but we feel guilty holding everyone back.

One glitch happens mid-week when we need to wash and prep all the syringes again. We realize we can't use the tap water to clean everything out, so we request a bowl but are denied one for fear of contagion. Instead, we are given clean garbage bags and line the sink with them before filling them with bottled water. A bit of extra mess and bother, but it gets the job done.

Amazingly, the trip goes without a health hitch for the youngest two. We cross the borders easily, and aside from one guard confiscating one of our three ice packs for some unknown reason, we arrive at our sunny destination and then home again to the snow: safe and sound, grateful for terrific memories of a week well spent.

Cystinosis didn't take a vacation – but it didn't wreck ours, either! And that is a beach-day win!

IN SICKNESS AND IN HEALTH

By Todd Braye

BLACKIE, ALBERTA, CANADA

I met her yesterday, or so it seems, but our paths crossed providentially in 1993. My seminary degree program required an internship on a church staff, and so I found myself in a small Canadian city in northern Alberta. The sands of time have blown, but one of my earliest recollections of her will never be buried. Upon a visit to the local shopping mall, a young, petite sales associate sold me a sweater. It had a soft purple tone, one of the colors she would choose for our future wedding.

It's beyond amazing to think that it was some 25 years ago. We were young, she was fun, and I was, well, eventually smitten. It was far from love at first sight, but our friendship would soon surprise us with romantic affections. I asked her to marry me in the fall of 1994, and before spring turned to summer in 1995 we covenanted to love each other "until death do us part." Faithfulness to each other was, and is, important to us. We would not sever this bond; only death could tear us apart. That much, it seemed to us, was easy. We desired very much to grow old together. But little did we know what that meant. Arguably, the words all too often pass over the lips with little if any weight, but oh how much would we learn about "in sickness, or in health!" However romantic the phrase sounds, the reality is anything but, especially when chronic disease increasingly shapes a couple's life.

When cystinosis wins another battle for my body, Bev is always there. A bone-chilling day in January 2013 was the first time. Somewhere between the parkade and the genetics clinic, it became obvious to both of us that I needed a wheelchair. I could no longer walk the distance. After finding one, I reluctantly sat. Bev pushed. It was humiliating, diminishing and defeating. As I sat, I remember choking back tears, feeling sorry for my wife; she deserved better. It was a difficult day for us both. But she was with me. Some run away. Some do not. Bev has not.

Confessedly, life has been far from easy. As cystinosis advances, my abilities – and thus my ability to enjoy life – decrease. Mobility issues, the constant medication regime, unrelenting trips to various specialists and lab tests, frequent frustration with fatigue, decreasing vocal ability, a keen sense of mortality, regular fights with my CPAP mask, and financial stresses are not the worst things, however. Though inconvenient, somewhat distracting and deeply sobering, I, we, have learned, and are learning, to deal with them. The toughest, most difficult reality I fight is muscle-wasting. Much ink is needed to explain, but it's enough to say I most miss being able to do the simple things, like go for walks in the rain, or embrace my wife. I need her help to hug her. So, one by one, she lifts my arms to her shoulders. When I finally pull her towards me, nothing is more heartbreaking and yet, at the very same time, heartwarming. But that's life, right? Who can deny that bitter and sweet kiss?

Perhaps bittersweet is the best way to describe our life together. Like any marriage, we have our good days and our bad, albeit both informed by my thorn in my flesh. The temptation to cave is often strong, but I refuse to give up. Bitter is the thorn; I despise it. But sweet is my Bev; I love her. By God's grace, we are doing it. We are growing old together, "'Til death do us part."

BY GOD'S GRACE, WE ARE DOING IT. WE ARE GROWING OLD TOGETHER

Unity Brings a Power for Change

By Josefin and Robert Kuhnel, Isabella's parents
SATILA, SWEDEN

We still remember the day Isabella got her diagnosis. The whole world just fell apart. The sadness and the helplessness we felt were huge. It is hard not to think about it – like the elephant in the room that you attempt to ignore. We tried to live a normal life but we didn't know how. We were so alone with no one to talk to, no one to ask for help.

We also remember the day that, as we were staring at the computer, we got big smiles on our faces when we found the Cystinosis Research Foundation and learned about their upcoming Day of Hope conference. Robert and I fell in each other's arms and cried the first tears of hope. We were going to the conference and actually had all of our bags packed and ready a whole month before our trip.

Going from the loneliness of being the only ones we knew with cystinosis, to meeting a whole community full of love, was life-changing. The community taught us about cystinosis and gave us hope for a bright future. We had hope not only for Isabella but for our family as a whole.

Going to the CRF Day of Hope is the highlight of the year. It's like an annual family reunion.

We believe that being in closer contact with people who are going through the same thing is extremely important. We have met families from all over – people in Sweden and Norway that we now have a beautiful connection with. Together, we're starting a foundation and a network in the Nordics that will help others living with cystinosis to get the same support and family feeling that we have felt because of CRF.

It's amazing how it's possible to create something so beautiful

GOING TO THE CRF DAY OF HOPE IS THE HIGHLIGHT OF THE YEAR. IT'S LIKE AN ANNUAL FAMILY REUNION.

and strong out of a heartbreaking disease, like cystinosis. The cystinosis families from all over the world have grown stronger together. We are so excited to be joining the conference along with three other families from the Nordic countries this year.

Isabella is turning nine in September and she appreciates this event more and more each year. She had met new friends and feels like she is "like everyone else" when she is at the conference. The only struggle for Isabella is the language barrier, but she's starting to learn English at school. Our goal has always been for Isabella to have a big cystinosis family to support her when she hits her teens.

Rare Disease Foundation in Sweden had a conference for families with cystinosis last year. At that conference we met another family with a boy Isabella's age. The two of them hit it off from the beginning, now they will meet again at the Day of Hope conference and have days of fun and sun together.

As parents, Isabella's disease has brought us so much life wisdom. Because of cystinosis we really know what to appreciate and what's important in life. What is most important though, is that as parents we can be strong in our daily life but as a community working together, we can change the future for a lot of wonderful people.

Hearts for Hadley

BIG HEARTS RAISE $100K FOR CYSTINOSIS RESEARCH

By Marcu Alexander, Hadley's mom

BOISE, IDAHO

The fourth annual Hearts for Hadley benefit took place on September 15, 2017 at Jack's Urban Meeting Place (JUMP) in downtown Boise, Idaho. The inspiring event raised $100,000 for cystinosis research.

The event welcomed more than 300 guests who were greeted by 7-year-old Hadley and big sister, Stella. Upon check-in, all attendees were gifted a Hearts for Hadley trucker hat generously donated by Citywide Home Loans.

Guests snacked on hors d'oeuvres while trying their luck on more than 20 raffle items. Some of the generously donated items included a three-night stay in Sun Valley, tickets to the Idaho Shakespeare Festival, original art, and a hand-crafted dollhouse made with love by Hadley's great aunt, Cherie. A wine wall was added this year and was a big hit! A limited number of tickets were available, and for $25 you were guaranteed a bottle of wine with the chance of getting a bottle valued up to $100.

The evening included a delicious menu specially created for Hearts for Hadley by local chef Jered Couch. A lively dessert auction took place after dinner and raised $10,700. The dessert auction has quickly become one of the favorite parts of the night and always sparks friendly competition among the crowd. The desserts are spectacular and could easily be mistaken as professionally made!

JJ Astorquia, long-time friend of the Alexander family, emceed the evening and acted as co-auctioneer. Donning his signature red pants and special "Uncle JJ" hat, JJ kept the crowd entertained and energized. Local TV news personality Mark Johnson joined JJ on stage as auctioneer during the live auction. The live auction was extra exciting this year! A weeklong guided rafting trip on the Middle Fork, a week in Mexico, and a puppy were just a few of the incredible items. The couple who won the puppy named her Hadley! A beautiful Prada bag made its appearance once again and to date has raised over $23,000 for CRF. It has been purchased and given back four times already. The winner of the bag this year immediately turned around and handed it to Hadley. Once again it will go up for auction in 2018!

This year's event was exceptional because of some special guests. Teresa, Patrick and Jenna Partington traveled from their home in Sacramento to support Hearts for Hadley. Patrick and Jenna are 13-year-old twins both living with cystinosis. The Partingtons are the first people the Alexanders connected with after Hadley's diagnosis at 18 months old. They have become great friends over the years and spend time with each other each summer at the lake in McCall, Idaho.

The outpouring of love and support by friends, family and the community of Boise is overwhelming! The Hearts for Hadley benefit continues to grow and help spread awareness for cystinosis. Planning is already under way for the fifth annual event this September.

Hearts for Hadley

H4H 2017

4th ANNUAL

HEARTS FOR HADLEY BENEFIT

1 NIGHT

15th OF SEPTEMBER

300 GUESTS

TONS OF AUCTION ITEMS

THE PRADA BAG, AGAIN!

$100,000 RAISED FOR CYSTINOSIS RESEARCH

IN HONOR OF 7-YEAR-OLD HADLEY

GROWING UP

IT'S OUR DIFFERENCES THAT MAKE US BEAUTIFUL

It seems like only yesterday that we were making arrangements for Olivia to have her G-tube put in, and here we are almost seven years later watching Olivia transition to swallowing pills and the possibility of finally removing her gastronomy tube.

I thought the day would never come. We have spent countless hours filling syringes, opening capsules and worst of all, cleaning the syringes. One day, Olivia randomly decided that she wanted to try swallowing pills after watching me take my morning vitamins. I was shocked at how easy it was for her to swallow one pill, and now she swallows them by the handful: the quicker the better. Olivia desperately wants to achieve her goal to rid herself of the G-tube in the near future. She is striving to prove that she can swallow all pills and eat a balanced diet before that happens. She still takes one round of vitamins via G-tube in the middle of the night, along with bolus feeds throughout the day. Her G-tube has been a lifesaver ensuring proper nutrition.

Olivia has taken on a huge part of her self-care, as well as the care of her overnight diaper changes and her own laundry care. It's hard watching her be so responsible because she is only 8 years old, but we know that in the long run these are the skills that will set her up for an independent life.

Our biggest struggle on Olivia's cystinosis path has been how to explain to her that she is different. She is starting to notice the things that she has to do daily compared to her sister and friends who do not. Although we are a very public family, we have been extremely protective of her innocence. I never anticipated how challenging it would be to look her in the eyes and tell her that she is different, especially in a world where we all want to fit in. There is no manual on how to tell her she is different without scaring her. We have explained to her that we are all different, and what makes you different makes you beautiful and the person you are.

Olivia is extremely compassionate and has a love for everyone. We recently found out that her sister Harper has some food allergies. One day while making lunch for the girls, Olivia looked me in the eye

and said, "I'm sorry mom that Harper can't have cheese. It's not fair that dairy makes Harper sick." I literally had to fight back tears realizing the compassion Olivia has for her sister's situation and how she doesn't view her own life as negative. Does Olivia wish she did not have to take all her vitamins? Yes, absolutely, but mostly because she hates being pulled away from her activities and playtime. She is still oblivious to the fact that her vitamins are what make her tummy hurt. We dread the day she makes this correlation.

We have learned to take one day at a time and celebrate the milestones. I can't believe how far she has come in seven years and how much we have grown as a family. Cystinosis has made us even stronger. We value the little things and have learned not to take things for granted. Olivia is the happiest little girl and one of the best things that has come from cystinosis is that she has a role model, Tina Flerchinger, who also has cystinosis. If Olivia is about to try a new food she always asks, "Does Tina eat this?" Our obvious answer is always, "Yes, she does." We are confident that Olivia will have a bright future, remain happy and become healthy.

OLIVIA HAS TAKEN ON A HUGE PART OF HER SELF-CARE

By Erin Little, Olivia's mom

PORT ELGIN, ONTARIO, CANADA

Showing Support
Through Collaboration and Networking

It's rare to have a disease that affects only 500 people in the United States. It might be even more rare to have an organization dedicate two annual events to support the 10-year-old daughter of an employee with a disease most people have never heard of. After Morgan Peachman's mom, Jen, had the opportunity to share a video shedding some light into the daily struggles of dealing with cystinosis on stage for 300 people in Nashville, the support provided to the Cystinosis Research Foundation in Morgan's honor has been absolutely tremendous!

Jen Peachman is vice president of member engagement at The Mortgage Collaborative, the nation's only independent cooperative network in the mortgage industry, boasting more than 200 lenders, members, and preferred partners. After the founders and board of directors of The Mortgage Collaborative (TMC) learned of Morgan's disease and the Peachman family's annual Mulligans Fore Morgan Charity Golf Tournament in 2017, the support TMC has shown for Jen, Morgan and the Cystinosis Research Foundation is a testament to the power of their network.

At their summer conference last August, TMC's Vice Chairman and President David G. Kittle, CMB, welcomed Jen on stage to discuss her daughter Morgan's struggles with cystinosis, to share information about the complexities of the disease, and her support of the Cystinosis Research Foundation. Kittle called upon the board of directors, employees, members, and guests in attendance to show their support of Mulligans Fore Morgan. On stage, he declared that beginning in 2018, the golf tournaments at TMC's bi-annual conferences would be charitable events – an extension of Mulligans Fore Morgan.

"NOTHING GIVES ME MORE PLEASURE THAN TO HELP MORGAN IN HER JOURNEY TO BE HEALED. SHE HAS THE FULL SUPPORT OF MY PARTNERS AND THE ENTIRE TMC FAMILY. WE HAVE MORGAN'S BACK!"

David G. Kittle, CMB,
Vice Chairman and President
The Mortgage Collaborative

TMC's 2018 winter conference kicked off on February 11th with 44 golfers teeing off at The Grand Golf Course in Del Mar, California, for the organization's first Mulligans Fore Morgan Charity Golf Tournament. Although Morgan couldn't join in person, her spirit was present. "Team Peach" even sported matching peach golf shirts to show their support of Morgan from afar. As of March, over $12,000 has been donated to the Cystinosis Research Foundation by friends of The Mortgage Collaborative as a result of the outing.

In February, Peachman and Kittle provided an update on TMC's support of the Cystinosis Research Foundation at their 2018 Winter Conference in Del Mar, California. Peachman was also joined on stage for a special check presentation by representatives from three extremely generous members of the cooperative, Success Mortgage Partners, Centennial Lending Group and People's United Bank.

The Mortgage Collaborative will continue its support of the Cystinosis Research Foundation by hosting another Mulligans Fore Morgan Charity Golf Outing in Chicago this August. Through support from TMC's network, the Peachman family's hope to find a cure for Morgan grows stronger. Through networking and collaboration, we find strength in numbers. We're stronger together.

Double-Digits & Double the Donations

By Jamie and Jen Peachman, Morgan's parents
AVON LAKE, OHIO

Our beautiful daughter, Morgan, turned 10 last year. Shortly after her big double-digit birthday, we held our family's 3rd Annual Mulligans Fore Morgan Charity Golf Tournament in Avon, Ohio. We're proud to share that we've more than doubled the donations made in Morgan's honor last year – totaling **$27,708** raised for the Cystinosis Research Foundation.

In addition to having a record-breaking year for our family's fundraiser, in the past year we've visited four states and attended eight events with our extended "cystinosis family."

We're already getting ready for the 4th Annual Mulligans Fore Morgan golf tournament.

Join us Sunday, September 16, 2018 at Bob-O-Link golf course!

BY STEPHEN JENKINS, MD

HELP! MY KID STILL

Cystinosis is hard. No one would say otherwise. But most people affected by cystinosis reach equilibrium, a "new normal," where they do what they have to do to keep going every day. Ashton and I got used to the frequent medications, the tube feeds, the vomiting, and all the other facets of cystinosis. We found work-arounds for most things and figured out how to lead mostly normal lives. The one thing I feel like we have not been able to conquer, however, is bed-wetting.

The first manifestation of cystinosis is Fanconi syndrome, which is the inability to reabsorb fluids, electrolytes, amino acids, and proteins from the urine, caused by injury to the tubular system in the kidneys. Fanconi syndrome leads to polyuria, or excessive urination. Some kids pee more than others, but all kids with cystinosis pee more than average.

We have two boys with cystinosis. Lars, our second child, was diagnosed shortly after birth and was started on cysteamine therapy right away. Thankfully, he has not developed full blown Fanconi syndrome yet, and he pees about as much as a kid without cystinosis.

Samuel, our first child, was diagnosed at the age of 1 and had pretty significant Fanconi syndrome by that time. He is now 8 years old, and the urination has not slowed down. If anything, it's gotten worse. For the most part he can manage this during the day, although I still have to remind him to go to the restroom when he starts dancing around. But we've all given up on night time. No matter what we do, every morning is a drenched mess. He doesn't seem to mind all that much, at least right now.

Sam goes to bed around 8 p.m. He wears a pull-up diaper with a poise pad inside. We've found that in males, at least, the poise pad works better if worn horizontally across the top of the pull-up. Because of male anatomy, urine tends to shoot upward, and the pad soaks up more of it in this position. He sleeps on a long bed pad, with a large fleece blanket on top. We've given up on regular sheets under the bed pad because he soaks through more often than not. The mattress has a waterproof liner on it. He drinks about two large bottles of water a night, in addition to 500 ml of formula from his tube feeds. By morning, his blanket, bed pad and pillow case are soaked, in addition to whatever stuffed animal he snuck into bed when we weren't looking. Everything goes into the washing machine first thing in the morning, and he hops in the bath. This is how every day starts, no matter what is going on in our lives. This is one of the reasons that in the eight years since Sam joined our family, we've only left him with someone else overnight once. It's the reason we rarely go camping, and sleeping in hotels can be stressful.

EVERYTHING GOES INTO THE WASHING MACHINE FIRST THING IN THE MORNING, AND HE HOPS IN THE BATH.

I've felt pretty defeated on this issue. We live in Utah, one of the outdoor capitals of the world, and I always dreamed I'd take my boys on long backpacking trips. Until we beat this bed-wetting thing, however, I don't think that's going to happen.

So I reached out on Facebook in the CRF Cystinosis Community Connection page for advice. Here's what I learned from other parents and patients dealing with this problem:

Many people recommend specific brands of diapers. All-Through-the-Night (ATN) diapers are supposed to have excellent absorption (think astronaut diapers). They can be ordered online through Home Delivery Incontinence Services (www.hdis.com). Other parents recommended Huggies night time pull-ups or Bambo diapers.

Some parents place a larger size diaper over the correct size diaper. Many use one size up and then place a variety of liners and pads in the diaper, including Poise pads, Sposie diaper booster pads, or All-Through-the-Night pads.

Many parents put disposable chucks on the bed, which saves you the trouble of laundry. We use disposable pads when we travel, specifically the kind with adhesive on the back so the pads don't move around at night. Others use washable pads, which is a good investment if you don't mind doing laundry every day. These come in different sizes, and the longest we've found is about 4 feet. Many parents also use waterproof mattress covers, which I highly recommend!

Many parents wake up to change their child's diaper during the night. We used to do that, until Sam started soaking out even before 10 p.m., and then it was like, what's the point? Others get their kids up one or two times a night to go, often the same time they do medication doses. We've tried it with Sam multiple times, but

WETS THE BED!

we usually give up after about a week. He just wants to sleep!

Multiple parents mentioned indomethacin. This is a nonsteroidal anti-inflammatory, related to ibuprofen, which reduces blood flow to the kidneys. One effect of this is to reduce urine output. For many children this works incredibly well. We started it for Sam over a year ago. It didn't seem to affect the volume of urine, but it has helped reduce his electrolyte losses. This is something that should only be used in consultation with a nephrologist, since it can cause worsening of the glomerular filtration rate (GFR) and requires careful monitoring.

Another medication that was mentioned was imipramine. This drug was originally designed as an antidepressant, but it has the side effect of dry mouth and urinary retention. This drug is commonly used in healthy children with bed-wetting. It should be used with caution in kids with cystinosis, however, because urinary retention in setting of polyuria can cause increased pressure in the bladder and the kidneys, which can cause permanent damage over time.

One parent mentioned that their child uses a bedside urinal, like those used in the hospital. It's basically a jug with a handle. Obviously this works best with males. Another parent said she got one of these for her child, but he doesn't wake up enough to use it. I've thought about using this for Sam (in fact we used to have one in the car) but I'm afraid he would tip it over, and then I'd be cleaning up a liter of urine out of the carpet! One adult with cystinosis said she had a bedside potty next to her bed. This might be a safer option, if I could get Sam to sit down when he pees.

Multiple parents mentioned using alarms. Some found it helpful, while some kids "are not a fan." I think these have the potential to work for many kids, and it's worth a shot. There are a lot of different options out there, of varying quality and expense, so this is something I would need to research more.

One parent said they restricted water intake at night. This is a technique commonly used in healthy children with bed-wetting, as well as older adults. I would consult your nephrologist before using this method, since children with cystinosis should usually have access to water so they can self-regulate and prevent dehydration. This is especially true if you are taking an ACE inhibitor, like enalapril or lisinopril, an angiotensin receptor blocker, like losartan, or indomethacin.

I think the biggest thing I took away from everybody's advice is that even though we are all affected by cystinosis, not every child or adult with cystinosis is the same. Some will stop wetting the bed by 5 years old, while other may have issues into their teen and even adult years. Some children decide on their own that they don't want to wear diapers anymore, and some children would rather get a good night of uninterrupted sleep, even if it means waking up drenched. While there is no "one-size-fits-all" strategy for bed-wetting and cystinosis, I appreciated everybody's willingness to help and to share their personal experiences. I think that is what makes our community special.

If you have other thoughts about cystinosis and bed-wetting, or you have other topics you'd like to read about in the future, send me an e-mail at stephenlars@gmail.com.

WE ARE Growing

In 2017, donors from the United States and around the world helped CRF raise more than $5.3 million for cystinosis research.

NORWAY

DENIS LILLAND
$565

BJORN ROTH
$2,845

AUSTRALIA

ERIK PEKLI
$6,123

CANADA

#TEAMSOPHIE
SOPHIE BETOURNAY
$30,525

EVA BILODEAU
$5,315

ANDREW CUNNINGHAM
$17,140

Seth's Circle of Hope
SETH DEBRYUN
$20,220

AMANDA KUEPFER
$4,777

MaryLynn's Road to a Cure
MARYLYNN LEPACK
$2,845

OLIVIA LITTLE
$183,896

ALIYAH AND MADELYN WALKER
$1,420

ABBI MONAGHAN
$13,165

KATHLEEN ROBERTS
$5,300

GABRIELLE STRAUSS
$13,989

Together

Your generosity continues to give hope to those with cystinosis and their families. That hope unites us and lights our way on the journey to better treatments and a cure.

UNITED STATES

HADLEY ALEXANDER
$123,338

LILY BEAUREGARD
$915

CHASE CHODAKOWSKY
$200

JOSHUA CLARKE
$15,273

BAILEY DEDIO
$5,089

TANNER EDWARDS
$1,555

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$67,444

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$14,375

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$3,880

CHARLOTTE COE
$1,280

BROOKE EMERSON
$20,415

TYLER JOYNT
$2,000

LANDON HARTZ
$24,393

MARY HEAD
$20,874

SAM AND LARS JENKINS
$3,929

JAKE KRAHE
$124,338

KENZIE LAWATSCH
$10,774

LOLA LONG
$60,230

PRESTON LUKE
$12,578

JENNA AND PATRICK PARTINGTON
$254,435

MORGAN PEACHMAN
$27,708

HENRY STURGIS
$200,798

KATIE KLOETE
$1,600

PRESTON LUKE AND BRANDON WALDRON FAMILIES
$1,160

KEEGAN MANZ
$9,481

AIDAN O'LEARY
$41,280

EMMA GRACE SUETTA
$2,913

BRADEN AND DAX TYNER
$4,080

FACEBOOK FRIENDS
$13,565

CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

2017 CRF *Research Grants Funded*

CYSTINOSIS RESEARCH FOUNDATION **GRANTS SUPPORT FOR SCIENTIFIC STUDIES ON *CELL FUNCTION, NEW TREATMENTS* AND *THE QUEST FOR A CURE***

ROLE OF MITOCHONDRIA IN NEPHROPATHIC CYSTINOSIS: THE CONTROL OF CYCLIC AMP

Francesco Bellomo, PhD, ***Principal Investigator***
Domenico De Rasmo, PhD, ***Co-Principal Investigator***
Bambino Gesù Children's Hospital – IRCCS, Rome, Italy
National Research Council (CNR), Bari, Italy

$150,150.00 – Eighteen-month grant
(February 1, 2018 – July 31, 2019)

IMPROVEMENT OF CELLULAR FUNCTION THROUGH CHAPERONE-MEDIATED AUTOPHAGY AND CELLULAR TRAFFICKING IN CYSTINOSIS

Sergio Catz, PhD, ***Research Mentor***
Jinzhong Zhang, PhD, ***Research Fellow***
The Scripps Research Institute, La Jolla, California

$75,000.00 – One-year grant
(September 15, 2017 – September 14, 2018)

MECHANISM OF BONE MARROW STEM CELL-MEDIATED THERAPY IN THE MOUSE MODEL OF CYSTINOSIS

Stéphanie Cherqui, PhD, ***Principal Investigator***
University of California, San Diego, La Jolla, California

$345,829.00 – Two-year grant
(September 1, 2017 – August 31, 2019)

INTRA-DERMAL IMAGING OF SUBJECTS WITH CYSTINOSIS USING CONFOCAL MICROSCOPY

Stéphanie Cherqui, PhD, ***Principal Investigator***
Ranjan Dohil, MD, ***Co-Principal Investigator***
Magdalene Dohil, MD, ***Co-Principal Investigator***
University of California, San Diego, La Jolla, California

Corinne Antignac, MD, PhD,
Co-Principal Investigator
Imagine Institute (Inserm U1163), Paris, France

$134,061.00 – One-year grant
(February 1, 2018 – January 31, 2019)

CLINICAL TRIAL READINESS FOR DISTAL MYOPATHY IN NEPHROPATHIC CYSTINOSIS (DMNC)

Florian Eichler, MD, ***Principal Investigator***
Reza Seyedsadjadi, MD, ***Co-Principal Investigator***
Massachusetts General Hospital, Boston, Massachusetts

$177,132.00 – One-year grant
(September 1, 2017 – August 31, 2018)

DEVELOPMENT OF A TOPICAL, CONTROLLED RELEASE CYSTEAMINE EYE DROP

Morgan Fedorchak, PhD, ***Principal Investigator***
Kanwal Nichal, MD, FRCO, ***Co-Principal Investigator***
University of Pittsburgh School of Medicine,
Pittsburgh, Pennsylvania

$163,819.00 – One-year grant
(September 1, 2017 – August 31, 2018)

TOTAL 2017 GRANTS FUNDED:

$2.46 million

FUNDING 13 RESEARCH PROJECTS

DYNAMICS OF CYSTINOSIN

Bruno Gasnier, PhD, ***Research Mentor***
Rosella Conti, PhD, ***Research Fellow***
Paris Descartes University, Paris, France

$150,000.00 – Two-year fellowship
(March 26, 2018 – March 25, 2020)

ELX-02 THERAPY FOR CYSTINOSIS CAUSED BY CTNS NONSENSE MUTATIONS

Paul Goodyer, MD, ***Principal Investigator***
McGill University Health Centre, Montreal, Quebec, Canada

$150,000.00 – Two-year grant
(September 1, 2017 – August 31, 2019)

NEWBORN SCREENING FOR CYSTINOSIS

Sihoun Hahn, MD, PhD, ***Principal Investigator***
Seattle Children's Research Institute, Seattle, Washington

$234,458.00 – Two-year grant
(September 1, 2017 – August 31, 2019)

AUTOPHAGIC LYSOSOMAL REFORMATION AND LIPID SIGNALING IN NEPHROPATHIC CYSTINOSIS

Alessandro Luciani, PhD, ***Principal Investigator***
Olivier Devuyst, MD, PhD, ***Co-Principal Investigator***
University of Zurich, Switzerland

$240,000.00 – Three-year grant
(October 1, 2017 – September 30, 2020)

IMPACT OF LEPTIN SIGNALING ON SKELETAL INTEGRITY AND GROWTH IN INFANTILE NEPHROPATHIC CYSTINOSIS

Robert Mak, MD, PhD, ***Principal Investigator***
University of California, San Diego, La Jolla, California

$167,177.00 – One-year grant
(September 1, 2017 – August 31, 2018)

IMPACT OF NLRP3 INFLAMMASOME SIGNALING ON SKELETAL INTEGRITY AND GROWTH IN INFANTILE NEPHROPATHIC CYSTINOSIS

Robert Mak, MD, PhD, ***Principal Investigator***
Harold Hoffman, MD, ***Co-Principal Investigator***
University of California, San Diego, La Jolla, California

$379,678.00 – Two-year grant
(February 1, 2018 – January 31, 2020)

SLEEP DISORDERS AND MEMORY IN ADULTS WITH CYSTINOSIS AND CHRONIC RENAL DISEASE

Doris Trauner, MD, ***Principal Investigator***
Atul Malhotra, MD, ***Co-Principal Investigator***
University of California, San Diego, La Jolla, California

$97,229.00 – One-year grant
(February 1, 2018 – January 31, 2019)

CELLULAR AND /OR MOLECULAR STUDES OF THE PATHOGENESIS OF CYSTINOSIS

43 GRANTS

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

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

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

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

Liang Feng, PhD
STANFORD UNIVERSITY, STANFORD, CALIFORNIA

Bruno Gasnier, PhD
Rossella Conti, PhD
PARIS DESCARTES UNIVERSITY, PARIS, FRANCE

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

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

Alessandro Luciani, PhD
UNIVERSITY OF ZURICH, 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
IMAGINE INSTITUTE, PARIS, FRANCE

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

STEM CELLS AND GENE THERAPY: BONE MARROW STEM CELLS, INDUCED PLURIPOTENT STEM CELLS, GENE THERAPY AND GENE EDITING

29 GRANTS

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

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

Paul Goodyer, MD
MONTREAL CHILDREN'S HOSPITAL, QUEBEC, CANADA

Patrick Harrison, PhD
UNIVERSITY COLLEGE CORK, IRELAND

Vasiliki Kalatzis, PhD
INSTITUTE GÉNÉTIQUE MOLÉCULAIRE MONTPELLIER, MONTPELLIER, FRANCE

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

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

CURE CYSTINOSIS INTERNATIONAL REGISTRY (CCIR)

1 GRANT

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

GENETIC ANALYSES 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

CYSTINE MEASUREMENT AND CYSTEAMINE TOXICITY STUDY

9 GRANTS

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

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

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

THYROID

1 GRANT

Pierre Courtoy, MD, PhD
DE DUVE INSTITUTE, UNIVERSITÉ CATHOLIQUE DE LOUVAIN, BRUSSELS, BELGIUM

KIDNEY RESEARCH

18 GRANTS

Robert Chevalier, MD
UNIVERSITY OF VIRGINIA, CHARLOTTESVILLE, VIRGINIA

Pierre Courtoy, MD, PhD
Christopher Pierreux, PhD
DE DUVE INSTITUTE, UNIVERSITÉ CATHOLIQUE DE LOUVAIN, BRUSSELS, BELGIUM

Olivier Devuyst, MD, PhD
UNIVERSITY OF ZURICH, INSTITUTE OF PHYSIOLOGY, ZURICH, SWITZERLAND

Allison Eddy, MD
BC CHILDREN'S HOSPITAL, VANCOUVER, BRITISH COLUMBIA, CANADA

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

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

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

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

NEW DRUG DISCOVERY CYSTEAMINE, NEW MEDICATIONS AND DEVICES

22 GRANTS

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

Pierre Courtoy, MD, PhD
DE DUVE INSTITUTE, UNIVERSITÉ CATHOLIQUE DE LOUVAIN, BRUSSELS, BELGIUM

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 Rega, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Paul Goodyer, MD
MONTREAL CHILDREN'S HOSPITAL, QUEBEC, CANADA

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

SKIN, MUSCLE AND BONE

8 GRANTS

Robert Ballotti, PhD
FACULTÉ DE MÉDECINE, NICE, FRANCE

Christine Chiaverini, MD, PhD
FACULTÉ DE MÉDECINE, NICE, FRANCE

Paul Grimm, MD
STANFORD UNIVERSITY SCHOOL OF MEDICINE, STANFORD, CALIFORNIA

Mary Leonard, MD, MSCE
STANFORD UNIVERSITY, STANFORD, CALIFORNIA

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

NEUROLOGICAL

15 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

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

EYE-CORNEAL CYSTINOSIS RESEARCH

8 GRANTS

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

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

Morgan Fedorchak, PhD
UNIVERSITY OF PITTSBURGH SCHOOL OF MEDICINE, PITTSBURGH, PENNSYLVANIA

Jennifer Simpson, MD
UNIVERSITY OF CALIFORNIA, IRVINE, GAVIN HERBERT EYE INSTITUTE, IRVINE, CALIFORNIA

Kang Zhang, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, SHILEY EYE INSTITUTE, LA JOLLA, CALIFORNIA

Total Grants Awarded:

$912,118

ROLE OF MITOCHONDRIA IN NEWPHOPATHIC CYSTINOSIS: THE CONTROL OF CYCLIC AMP

Francesco Bellomo, PhD, Principal Investigator
Domenico De Rasmo, PhD, Co-Principal Investigator
Bambino Gesù Children's Hospital – IRCCS, Rome, Italy
National Research Council (CNR), Bari, Italy

$150,150.00

18 MONTH GRANT
(February 1, 2018 – July 31, 2019)

INTRA-DERMAL IMAGING OF SUBJECTS WITH CYSTINOSIS USING CONFOCAL MICROSCOPY

Stéphanie Cherqui, PhD, Principal Investigator
Ranjan Dohil, MD, Co-Principal Investigator
Magdalene Dohil, MD, , Co-Principal Investigator
University of California, San Diego, La Jolla, California
Corinne Antignac, MD, PhD, Co-Principal Investigator
Imagine Institute (Inserm U1163), Paris, France

$134,061.00

ONE-YEAR GRANT
(February 1, 2018 – January 31, 2019)

DYNAMICS OF CYSTINOSIN

Bruno Gasnier, PhD, Research Mentor
Rosella Conti, PhD, PhD, Research Fellow
Paris Descartes University, Paris, France

$150,000.00

TWO-YEAR FELLOWSHIP
(March 26, 2018 – March 25, 2020)

IMPACT OF NLRP3 INFLAMMASOME SIGNALING ON SKELETAL INTEGRITY AND GROWTH IN INFANTILE NEPHROPATHIC CYSTINOSIS

Robert Mak, MD, PhD, Principal Investigator
Harold Hoffman, MD, Co-Principal Investigator
University of California, San Diego, La Jolla, California

$379,678.00

TWO-YEAR GRANT
(February 1, 2018 – January 31, 2020)

SLEEP DISORDERS AND MEMORY IN ADULTS WITH CYSTINOSIS AND CHRONIC RENAL DISEASE

Doris Trauner, MD, Principal Investigator
Atul Malhotra, MD, Co-Principal Investigator
The Scripps Research Institute, La Jolla, CA

$97,229.00

ONE-YEAR GRANT
(February 1, 2018 – January 31, 2019)

SEE LAY ABSTRACTS STARTING ON PAGE 75

WORKING TOGETHER TO FIND THE CURE

THANK YOU CANADA!

We are forever grateful to our Canadian cystinosis families who have worked in partnership with the Cystinosis Research Foundation (CRF) since 2009 to fund research. In fact, Canadian families have funded over $400,000 in research grants!

Canadians can send donations directly to CRF or they can contribute to CRF Scientific Review Board approved research studies through Canada Helps-Canadian Cystinosis Research Foundation Fund managed by the Aqueduct Foundation and administered by Cystinosis Awareness Research Effort (CARE). Through Canada Helps, CARE has created an efficient and effective fundraising process which allows Canadians to fund raise and ensure their donors receive a charitable tax receipt.

The Cystinosis Awareness and Research Effort and the Liv-A-Little Foundation have a strong working relationship with CRF. Erin Little (Liv-A-Little) is a CRF board of trustee member. Karen McCullagh, Crystal Walker and Chad Little are co-chairs of CARE and help guide the funding process.

Since 2003, CRF has funded 164 multi-year studies in 12 countries and is the largest fund provider of cystinosis research in the world. CRF has a scientific review board composed of world renowned cystinosis scientists and experts who review and recommend research applications for funding.

CRF and CARE would like to acknowledge the contributions of Jody and Trevor Strauss. They were instrumental in creating a focal point for Canadian families when they started Cystinosis Awareness Research Effort. Jody was an active member of CARE and the CRF board and Trevor held the role of President of CARE until November 2017. We are forever grateful for their leadership, commitment and contributions to the cystinosis community.

We are thankful to all of our Canadian families who are dedicated to finding better treatments and a cure. Together our two countries have united 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. Great things happen when countries work together!

A very special thank you to the following Canadian cystinosis families who have helped raise money and donated to find a cure for cystinosis:

- *Sophie Betournay*
- *Valéri Talbot and Eric Bilodeau*
- *Amanda and Dave Buck*
- *Monique and Don Carriere*
- *Sue and Pete Chatelain*
- *Karen McCullagh and Don Cunningham*
- *Kristen Murray and Nathan deBruyn*
- *Marthe and Rick Drolet*
- *Rachel and Mahlon Kuepfer*
- *Erin and Chad Little*
- *Fannie and Wayne Martin*
- *Katie and Terry Monaghan*
- *Susan and Peter Penner*
- *Marianne Sincennes and Daniel Picard*
- *Liz Ewart and Dan Roberts*
- *Jody and Trevor Strauss*
- *Diane and Elroy Wagler*
- *Crystal and Bob Walker*

Since 2016, Canadian families have directly funded grants through Aqueduct totaling $463,012.75

The following grant payments were recently paid by donations from Canadian families:

Liang Feng, PhD
STANFORD UNIVERSITY

Molecular mechanism of cystinosis
$37,500
Funded by Aqueduct Foundation

Paul Goodyer, MD
MCGILL UNIVERSITY HEALTH CENTRE

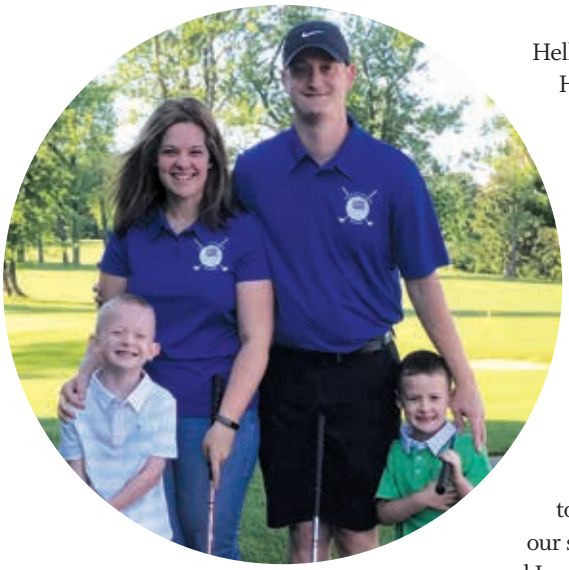
ELX-02 therapy for cystinosis caused by CTNS nonsense mutation
$37,500
Funded by Liv-A-Little Foundation

If you would like to learn more about how to fundraise in Canada, please contact CRF Board Member Erin Little (*ce.little@bmts.com*) or Karen McCullagh (*KCMccullagh@gmail.com*)

CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

Welcome New Board Member

LAUREN HARTZ



Hello! I live in Pittsburgh, Pennsylvania, and am the wife of Jimmy Hartz and mom to two boys, Landon and Jordan. Landon was diagnosed with cystinosis at the age of 14 months and was hospitalized for 28 days at the time of his diagnosis. While in the hospital, Jimmy and I connected with CRF Founder Nancy Stack and our fear and helplessness transformed into optimism and motivation. We planned our very first fundraiser and with help from family and friends, held an event for 300 people five months after being told that our son had cystinosis. Our first fundraiser has been followed by 11 more with two more on the calendar for 2018.

I took advantage of all of the doctors appointments, therapy appointments, time in my new role as a stay-at-home mom, and connecting with others in the cystinosis community to learn and to expand our support system in a way that helped our son to thrive even more. It does take a village to raise a child and I gathered whatever information I needed to from that village, to move forward.

As time went on and Landon's needs became more manageable, our family welcomed another little boy into our life and I began to explore who I was prior to being a mom. I identified some emotional and self-care needs that I had been neglecting and got to work on learning about myself, my grief, and how I connect with my husband, children, family, friends, and community.

I went back to work, part-time, armed with new knowledge and confidence. I was a mental health therapist prior to quitting my job and the need to do that work again became strong. I became licensed as a professional counselor in Pennsylvania and began working with children, teens and their families. Soon after going back to work I decided, with a gentle nudge from my husband, to start my own solo private practice. My niche, to empower women and young women to move through anxiety, depression and to go on a journey of self-awareness, acceptance and care felt like a natural fit.

I believe that my compilation of personal and professional experiences is a natural fit for the mission of the Cystinosis Research Foundation. In addition to fundraising, a personal connection with the cystinosis community, and work as a therapist, I have also had the experience of working as a research coordinator on several pharmaceutical research grants while in graduate school. This has provided me with an understanding of the research process and all that is involved.

Serving as a board member for this community is an honor that I cannot express adequately through words. Sharing our story has been very therapeutic for me throughout these last seven years and listening to your stories has taught me so much about the disease, about empathy, about resilience, about love, and about hope. Thank you!

CYSTINOSIS RESEARCH FOUNDATION
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Welcome New Board Member

NATALIE STACK

It is an honor to be considered for a board of trustee position for the Cystinosis Research Foundation.

I was born and raised in Corona del Mar, California. At the age of 7 months, I was diagnosed with cystinosis. My parents, Jeff and Nancy Stack, gave me my medicine religiously every day which I believe has helped me stay as healthy as possible.

Around the age of 10 or 11, I somehow figured out that I had an incurable disease. On the eve of my 12th birthday, my mom asked me what my birthday wish was; she told me I could wish for anything in the world. I was too shy to tell her aloud, so I scribbled my wish on a napkin. I wrote, "To have my disease go away forever." I finally showed my mom the napkin and, after reading the wish, my mom knew that she had to do something to save me and organized an event to raise money to find the cure. Soon after that fateful day when I made my wish, my mom and dad founded the Cystinosis Research Foundation hoping that one day I would be cured.

I just turned 27 years old and I live in Irvine, California. I received my bachelor's degree in psychology from Georgetown University in Washington, D.C. and my master's in social work from the University of Southern California. I have a passion for helping others and am interested in the nonprofit area. I hope to work with adoptive and foster care children in the near future.

Currently I work as a development manager for Taller San Jose Hope Builders, a nonprofit organization that equips disadvantaged young adults with the job-training and life skills needed to move out of poverty and onto a path of self-sufficiency. I assist with corporate and foundation relations which include: writing and managing grants, managing the volunteer program, and assisting with corporate sponsorship solicitations. I have excellent communication skills, I am detail-oriented, compassionate, reliable, and always ready to challenge myself.

I am thrilled to be a CRF board member. There are several areas where my talents and interests will benefit the foundation. I can assist with social media, I can communicate with other adults who have cystinosis, I will advocate for patients and their families, and I would like to help with the annual CRF Day of Hope family conference. I believe my background and my life experiences to date, will make me an excellent board member.

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.

TOGETHER, WE ARE one

1 PURPOSE.
1 JOURNEY.
1 CURE.

Mary Head — Rainier, Washington

Music For Mary

Music for Mary was held on February 24th at the Centralia Grand Hotel and Ballroom in Centralia, Washington. Friends, family and community all made the long drive to support Mary Head.

The evening started off with hors d'oeuvres, Italian sodas and a silent auction! Everyone then headed upstairs to the ballroom for a sit-down Italian dinner and music by the Head's favorite singer and band, Chris Anderson. He is a crowd pleaser with his amazing crooner style voice!

The Head's dear friend, Shannon Keizer, made the trip from Michigan to speak. Her speech was elegant, inspirational and informative about her journey as an adult with cystinosis!

J.R. and Melissa's friend, Matt Smith, did a great job as emcee for the night and auctioneer, Bob Kamstra, was hysterical when he took over the live auction and rallied the crowd!

The evening was a wonderful success for cystinosis research!

Hadley Alexander — Boise, Idaho

Hearts for Hadley Christmas Bus

Hearts for Hadley

The fourth annual Hearts for Hadley Benefit was a record-breaking success last year, due in part to some creative fundraising ideas. This past year, a friend of Ben and Marcu Alexander's proposed adding something new to the yearly auction, a Christmas costume party traveling by bus around Boise. The bus was generously donated by Boise Shuttle, owned and operated by Caldwell Transportation.

"Our friend Cody came up with the idea and just ran with it," said Marcu Alexander. "People couldn't sign up fast enough! It was the party of the season."

In total, 26 people filled the bus, all dressed in costumes, from Cindy Lou Who to Cousin Eddie from National Lampoon's "Christmas Vacation." The bus stopped at various bars and restaurants around town, which allowed the participants to share the story of Hearts for Hadley with the larger community.

"I was filled with so much pride seeing everyone dressed to the holiday nines and living it up while also raising money to help Hadley and everyone living with cystinosis," Marcu Alexander said. "The Christmas Bus raised $1,300 for Hearts for Hadley and is already booked for December 2018!"

TOGETHER, WE ARE one

Lola Long — Chaska, Minnesota

'Curl for the Cure' Gets Some Lift from Olympics

Lola's second "Curl for The Cure" was held on March 3, 2018 at the Chaska, Minnesota, Curling Center. We were pleased to have hosted a sold-out event and are extremely grateful for all of the friends and family who were able to attend. The curling competition was particularly popular this year — no doubt with some lift from the U.S. success at the Winter Olympics.

After the curling sessions had ended, we filled the event center with over 220 people. Dinner and drinks were served and everyone had the opportunity to bid on an incredible selection of silent auction items. Raffle items were available and a wine toss made possible by local wine shops and Miner Family Vineyards in Napa, California.

We would like to thank all of our sponsors for their gracious support as well as everyone in attendance. We feel so fortunate to have raised over $80,000 this year and to be able to share Lola's story, raise awareness for cystinosis and help fund research for a cure.

Internet

Facebook Funders Further the Quest for a Cure.

A huge thank you to all of our dedicated Facebook event organizers and donors! You have done an outstanding job raising funds for research. In 2017, you raised over $13,565 for CRF! Your donations make a difference and have helped us advance research that is focused on finding better treatments and a cure. Thank you for your commitment to CRF as we continue our quest for a cure together!

1 PURPOSE. 1 JOURNEY. 1 CURE.

Henry Sturgis — Sandpoint, Idaho

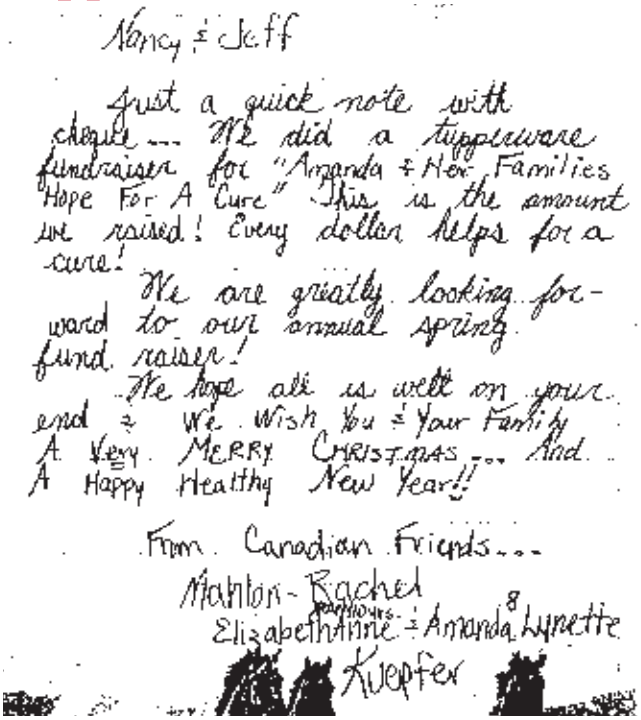
Swing for a Cure Raises Funds for Research

On September 29, 2017, Randi Lui, a longtime supporter of "24 Hours for Hank," held her first golf tournament at Stoneridge Golf Course in Blanchard, Idaho. With a nearly sold-out tournament she raised over $10,000 for cystinosis research! Several holes had entertaining games to play at the tee box to raise additional dollars and one hole had a chance to win a new car with a hole in one. Unfortunately, no one was lucky enough to win it this year. The tournament, held in honor of Henry Sturgis, was followed by an amazing awards dinner.

Sponsors included Global Credit Union, Six Stat Automotive, Jalapenos, Potlatch No. 1 Federal Credit Union, SandCreek Custom Wear, SimulStat, TraskBritt, Sayler Owens & Kerr, and Alpine Motors. Randi has already secured her date for this year's golf tournament, August 25, 2018.

Amanda Kuepfer — Ontario, Canada

Tupperware Raises $511



Nancy & Jeff

Just a quick note with cheque ... We did a tupperware fundraiser for "Amanda & Her Families Hope For A Cure" This is the amount we raised! Every dollar helps for a cure!

We are greatly looking forward to our annual spring fund raiser!

We hope all is well on your end & We Wish You & Your Family A Very MERRY CHRISTMAS ... And A Happy Healthy New Year!!

From Canadian Friends ...

Mahlon - Rachel
Elizabeth Anne & Amanda Lynette
Kuepfer

Amanda and her Family's HOPE FOR A CURE

TOGETHER, WE ARE one

Tina Flerchinger — Clarkston, Washington

Dealership Offers Ongoing Support

Tina's Hope for a Cure has been the recipient of Rogers Toyota's "Choose Your Charity" campaign for the third consecutive year. Rich and Ryan Rogers are known for their generous community involvement. They have been a big part of our success in raising awareness and funds for cystinosis research. For five consecutive years Rogers Toyota hosted our Wine, Stein & Dine event in their beautiful showroom in Lewiston, Idaho. We are appreciative of their continual support!

Katie Roy Kloete — Franklin, Tennessee

Cookin' for a Cure

We just submitted a $500 donation to cystinosis research! Rick and I hosted a chili cook-off recently. Voting was done by dollar donation. We brought in $440, but Rick and I rounded it up to $500 again, not a huge amount but our combined events this year (crawfish boil and chili cook-off) have raised a total of $1,600 for cystinosis research!

1 PURPOSE. 1 JOURNEY. 1 CURE.

Kaleb Lawshe — Charleston, South Carolina

Quinn's Fundraiser

Our cousin, Quinn Davis, is a beautiful young woman, both inside and out. She has an adorable 6-year-old son and will marry the love of her life in early June. Throughout our journey with cystinosis, we have learned one of the most important things; the support of family and friends is what keeps us going.

In honor of her own January birthday, Quinn surprised us by creating a fundraiser on Facebook in support of the Cystinosis Research Foundation (CRF). We were so overwhelmed with gratitude that someone would take the time out of their very busy schedule to put this together in honor of Kaleb.

Quinn's birthday goal was $200 and through the support of family and friends, she raised over $700 within just a few weeks. Quinn not only thought about Kaleb in creating the fundraiser, she also gave us a special gift – creating more awareness, and the funds for more research to bring us a step closer to finding a cure for cystinosis.

Thank you Quinn – we love you and are forever grateful!

Jenna and Patrick Partington — Sacramento, California

Memorial Donations Toward a Cure

The Partington Family and Jenna & Patrick's Foundation of Hope would like to thank the Pollacchi Family for selecting JPFH for memorial donations in honor of Leonard Pollacchi, who passed in February 2018. Leonard was a devoted husband to Mrs. Dona Pollacchi, a teacher at Holy Spirit School for over thirty years, and she was Jenna and Patrick's kindergarten teacher. Dona has had a tremendous, loving influence on Jenna and Patrick's primary school years, and we are humbled by the decision that she and Leonard made together before he passed. When she called to ask if donations might be made to JPFH in her husband's memory, she quoted Leonard's words: "I would like to know that contributions will be made for the study of cystinosis. I have had a long, healthy and happy life. I want Jenna, Patrick and others to have the same."

It Takes A Village

ACTIVITIES CALENDAR

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 1, 2018

LOTS OF LOVE FOR LANDON GOLF OUTING IN HONOR OF LANDON HARTZ

Blackhawk Golf Course, Beaver Falls, Pennsylvania

Month of August, 2018

PAINT THE TOWN PURPLE IN HONOR OF OLIVIA LITTLE

Saugeen Shores, Ontario, Canada

Sunday, August 12, 2018

KENZIE'S DRIVING FOR A CURE IN HONOR OF KENZIE LAWATSCH

Little River Country Club, Marinette, Wisconsin

For information: Shawn Lawatsch 715-587-4317

Saturday, August 18, 2018

TMC MULLIGANS FORE MORGAN CHARITY GOLF OUTING

Harborside International Golf Club, Chicago, Illinois

Saturday, September 8, 2018

7TH ANNUAL SWING, SHOOT AND LIV GOLF CLASSIC IN HONOR OF OLIVIA LITTLE

Saugeen Golf Club, Port Elgin, Ontario, Canada

For information: Erin.Little@livalittlefoundation.com

Saturday, September 15, 2018

5TH ANNUAL HEARTS FOR HADLEY - HADLEY ALEXANDER

JUMP, Boise, Idaho | *For information: Marcu Alexander, hearts4hadley@gmail.com*

Sunday, September 16, 2018

4TH ANNUAL MULLIGANS FORE MORGAN, MORGAN PEACHMAN

Bob-O-Link Golf Course, Avon, Ohio

For information: Jennifer Peachman, jennifer.peachman@gmail.com

Friday and Saturday, October 5-6, 2018

8TH ANNUAL SWING & BLING EVENT JENNA AND PATRICK PARTINGTON'S FOUNDATION OF HOPE

Swing Golf Event - Catta Verdera Country Club

Bling Gala Dinner - The Sawyer Hotel, Sacramento, California | *For information: www.jpfh.org*

Thursday-Saturday, March 28-30, 2019

CYSTINOSIS RESEARCH FOUNDATION DAY OF HOPE FAMILY CONFERENCE AND NATALIE'S WISH CELEBRATION

Island Hotel, Newport Beach, California

CYSTINOSIS RESEARCH FOUNDATION
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Role of Mitochondria in Nephropathic Cystinosis: The Control of Cyclic AMP

Francesco Bellomo, PhD, Principal Investigator
Domenico De Rasmo, PhD, Co-Principal Investigator
Bambino Gesù Children's Hospital – IRCCS, Rome, Italy
National Research Council (CNR), Bari, Italy

OBJECTIVE/RATIONALE:

Most lysosomal storage diseases show mitochondrial impairment. Lysosomal dysfunction, in fact, negatively affects mitochondrial function, and vice versa. Mitochondria have an important role in energy supply, calcium homeostasis, ROS balance and apoptosis, and tissues with high metabolic activity like liver, kidney and neuromuscular apparatus, result more susceptible to the mitochondrial damage. Recent findings show that cyclic AMP, one of the main second messengers of the intracellular signal transduction cascades, is compartmentalized into subcellular structures, including mitochondria, where it could exert several regulatory effects.

PROJECT DESCRIPTION:

The aim of our research project is to perform a functional and structural analysis of mitochondrial dynamics in different cellular models of cystinosis. We will investigate molecular mechanism of mitochondrial scission and fusion processes in two human models of cystinosis: conditioned immortalized proximal tubular cells of patient and CRISPR/Cas9 knock-out of CTNS gene in the human skeletal muscle cells, and in a murine model obtained by isolation of myoblasts from gastrocnemius muscle of CTNS-/- mice. Then, we will evaluate the significance of the cyclic AMP signal in mitochondrial bioenergetics via specific pharmacological treatments.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

While mitochondrial contribution in the pathophysiology of cystinosis is emerging, mechanistic details still need to be elucidated. Apoptosis and mitophagy are increased in cystinotic cells and these processes could be the initial trigger to the development of the clinical disease. Drugs and nutraceutics that modulate cyclic AMP levels could ameliorate mitochondrial bioenergetics, especially in those tissue with a high energy demand.

ANTICIPATED OUTCOME:

We expect to obtain further evidences that show mitochondrial impairment in cystinosis and clarify molecular mechanism underlying the apoptosis associated to the cystinotic phenotype. These findings could potentially be leading to identify a supplementary treatment for the kidney failure and distal myopathy observed in cystinosis.

Intra-dermal Imaging of Subjects with Cystinosis using Confocal Microscopy

Stéphanie Cherqui, PhD, *Principal Investigator*
Ranjan Dohil, MD, *Co-Principal Investigator*
Magdalene Dohil, MD, *Co-Principal Investigator*
Corinne Antignac, MD, PhD, *Co-Principal Investigator*
UNIVERSITY OF CALIFORNIA, SAN DIEGO

OBJECTIVE/RATIONALE:

This study intends to gather intra-dermal images of the skin from patients with cystinosis as a method to estimate the quantity of cystine crystals in this tissue compartment. The tissue cystine crystal count will be correlated with disease outcome and may correlate with long-term efficacy of treatment and compliance with therapy.

PROJECT DESCRIPTION:

At least 50 patients affected with cystinosis at various ages will be imaged to gather intra-dermal images of their skin and compared to healthy subjects using the confocal microscope (VivaScope® 3000). Cystine crystals will be quantified for each patient using an imaging software. Intra-dermal cystine crystal counts will be correlated with clinical outcomes such as glomerular filtration rate, thyroid function test, growth, and with treatment history (with cysteamine), especially posology of cysteamine and compliance to therapy. Genetic mutations for each patient, if not known, will be identified in the laboratory of Dr. Corinne Antignac.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This present study looks at an alternative method to monitoring the levels of cystine crystal accumulation in the skin and compares that relationship to the current method of blood cystine levels, and to potentially correlate these levels to treatment compliance, their health status (kidney function, endocrine function, etc.) and genetic mutations. The outcome of this study will be supportive of the future stem cell gene therapy clinical trial and may outline intra-dermal imaging as a supportive test in monitoring patients affected with cystinosis over time.

ANTICIPATED OUTCOME:

The outcome of this study will be supportive of the future stem cell gene therapy clinical trial and may outline intra-dermal imaging as a supportive test in monitoring patients affected with cystinosis over time.

Dynamics of Cystinosin

Bruno Gasnier, PhD, *Research Mentor*
Rosella Conti, PhD, *Research Fellow*
PARIS DESCARTES UNIVERSITY, FRANCE

OBJECTIVE/RATIONALE:

The aim of this project is to improve our understanding of cystinosin, the protein defective in cystinosis, at the molecular level. We know the main biochemical function of cystinosin – it exports the amino acid cystine from the lysosome – but we do not know how the protein works. Moreover, novel molecular functions of cystinosin have been suggested. This project aims to identify key molecular mechanisms of cystinosin using biochemical and biophysical approaches to analyze its functions in an accurate manner.

PROJECT DESCRIPTION:

Our research plan will focus on two critical, complementary mechanisms of cystinosin: first, its coupling to the ATP-powered proton pump (V-ATPase) of the lysosome, a major regulator of lysosomal function; second, the existence of structural changes.

The V-ATPase pumps protons into the lysosome, thus acidifying its content. This acidity in turn activates cystinosin and drives cystine out from the lysosome by co-transporting cystine with protons. In a previous study, we have identified the proton binding site involved in this mechanism. We will now examine how this proton binding site is regulated by cystine using mutagenesis and electrophysiological techniques.

As with other membrane transporters, cystinosin must undergo structural rearrangements to transport cystine through the lysosomal membrane. These structural changes have never been observed and they represent a technical challenge to characterize the structure of cystinosin by crystallographic techniques. We will reveal such structural changes using biochemical techniques, and attempt to freeze cystinosin in a specific conformation using site-directed mutagenesis.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Lysosomal accumulation of cystine is a hallmark of cystinosis and its reversal by cysteamine alleviates symptoms. However, cysteamine treatment does not cure the disease and some cellular defects are not rescued by cysteamine, suggesting the involvement of novel cystinosin functions unrelated to cystine transport in the pathology.

The characterization of structural changes and key amino acids associated with cystine transport and the identification of specific mutants disrupting these properties will provide accurate information and tools to characterize these novel cellular functions of cystinosin and their contribution to the pathology.

ANTICIPATED OUTCOME:

Our project will improve our understanding of the cystine transport function of cystinosin, and help the progression and interpretation of higher resolution studies performed at the atomic level. It will also provide the cystinosis research community with accurate information and tools (mutants) to investigate the cellular functions of cystinosin and the multiple facets of cystinosis pathology with high precision, and help to understand the limitations of the cysteamine treatment.

Impact of NLRP3 Inflammasome Signaling on Skeletal Integrity and Growth in Infantile Nephropathic Cystinosis

Robert Mak, MD, PhD, *Principal Investigator*
Harold Hoffman, MD, *Co-Principal Investigator*
UNIVERSITY OF CALIFORNIA, SAN DIEGO

OBJECTIVE/RATIONALE:

Bone disease and growth retardation are common in children with CKD (chronic kidney disease). Skeletal manifestations in patients with infantile nephropathic cystinosis (INC) were presumed to be similar to those described in CKD. However, there is a surprising lack of detailed information on the skeletal abnormalities in INC. The NLRP3 inflammasome may negatively impact skeletal integrity and growth in children with INC.

PROJECT DESCRIPTION:

We will delineate the role of the NLRP3 inflammasome in bone complications in a mouse model of INC, Ctns-/- , by using both a genetic and pharmaceutical approach. We propose the following specific aims: (1) We will compare the bone phenotype over the Ctns-/-NLRP3-/- and Ctns-/-IL-1ß-/- double knockout mice relative to Ctns-/- mice and wildtype control mice; (2) We will investigate the impact of IL-1ß targeted therapy on skeletal integrity and bone strength in Ctns-/- mice using Anakinra (IL-1 receptor antagonist).

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Skeletal manifestations described in INC patients included failure to thrive, rickets, pathological fractures and short stature, for which there are no effective treatments. This is an urgent unmet medical need. Funding from CRF is critical to develop innovative therapy for bone complications in patients with INC. Novel therapeutic approaches are required to ameliorate progressive destructive bone changes patients with INC, which often necessitate orthopedic surgery.

ANTICIPATED OUTCOME:

We hypothesize that activation of the NLRP3 inflammasome may negatively impact skeletal integrity, bone strength (fractures) and growth in children with INC. This pre-clinical study may form the basis of a clinical trial. Our ultimate goal is a clinical trial based on the results of this important translational study.

Sleep Disorders and Memory In Adults with Cystinosis and Chronic Renal Disease

Doris A. Trauner, MD, *Principal Investigator*
Atul Malhotra, MD, *Co-Principal Investigator*
UNIVERSITY OF CALIFORNIA, SAN DIEGO

OBJECTIVE/RATIONALE:

Many adults with cystinosis express concerns about their memory, but specific memory functions have not been previously evaluated in a group of adults with cystinosis. A recent questionnaire study involving 50 adults with cystinosis identified a high incidence of sleep complaints in these individuals, although specific details were not provided as part of that questionnaire. This study will examine the incidence, type, and severity of sleep disturbance in adults with cystinosis, as well as memory functions in those individuals, and the possible relationship between sleep disturbance and memory concerns in cystinosis. We will also study sleep and memory in adults with chronic renal disease from other causes to determine whether the sleep disturbance is specific to cystinosis or whether it is a general consequence of chronic renal disease. In either event, the results would have profound implications for increasing awareness, early diagnosis, and effective treatment of sleep disorders in young adults with these conditions, with the potential for improvement in quality of life and overall functioning.

PROJECT DESCRIPTION:

The study will involve travel to San Diego for an overnight sleep study in a sleep laboratory at the University of California, San Diego, as well as tests of attention and memory. We hope to recruit 20 adults with cystinosis and 10 adults with chronic renal disease of other causes. We will provide transportation arrangements to San Diego, hotel accommodations, and cover meal expenses while participating in the study.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

If sleep problems are identified in adults with cystinosis (or other chronic renal disease), treatments may be available that could improve quality of life. If sleep disturbances are found to be associated with memory impairments, treatment of the sleep disorder could significantly improve cognitive functioning and quality of life for adults with cystinosis and possibly for adults with chronic renal disease of other causes as well.

ANTICIPATED OUTCOME:

If the results of the study are positive, there is the potential to improve quality of life and possibly memory functioning in adults with cystinosis.

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2018

Call for Research Proposals

LOOKING AHEAD

Research is our Hope!

When Nancy and Jeff Stack established the Cystinosis Research Foundation 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 15 short years. Since its inception, CRF has raised more than $40 million with every dollar donated going directly to 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 2018 spring call for research proposals and fellowship grants. The grant awards will be announced in June 2018. Details and guidelines for the fall 2018 applications will be available September 1 online at the CRF website: w*ww.cystinosisresearch.org/research/for-researchers*.

In 2017, CRF issued 13 new grants for $2.46 million that brings us closer to better treatments and a cure. All research applications received by CRF are evaluated by CRF's Scientific Review Board (SRB) comprised of leading international experts in the field of cystinosis, on page 26. 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.

In 2010, CRF established the **Cure Cystinosis International Registry (CCIR)** to serve as a hub of information about cystinosis and its complications. Currently, CCIR has 576 registrants from 44 countries. The site, which includes a Professional Research Portal, is a critical resource for researchers and scientists who register to access and view de-identified, aggregate cystinosis patient information. The portal can be accessed at *www.cystinosisregistry.org*.

www.cystinosisresearch.org/research/for-researchers

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.

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Zoe R. Solsby
Vice President

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 $40 million with 100% of all your donations going to support cystinosis research.

EDUCATION

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

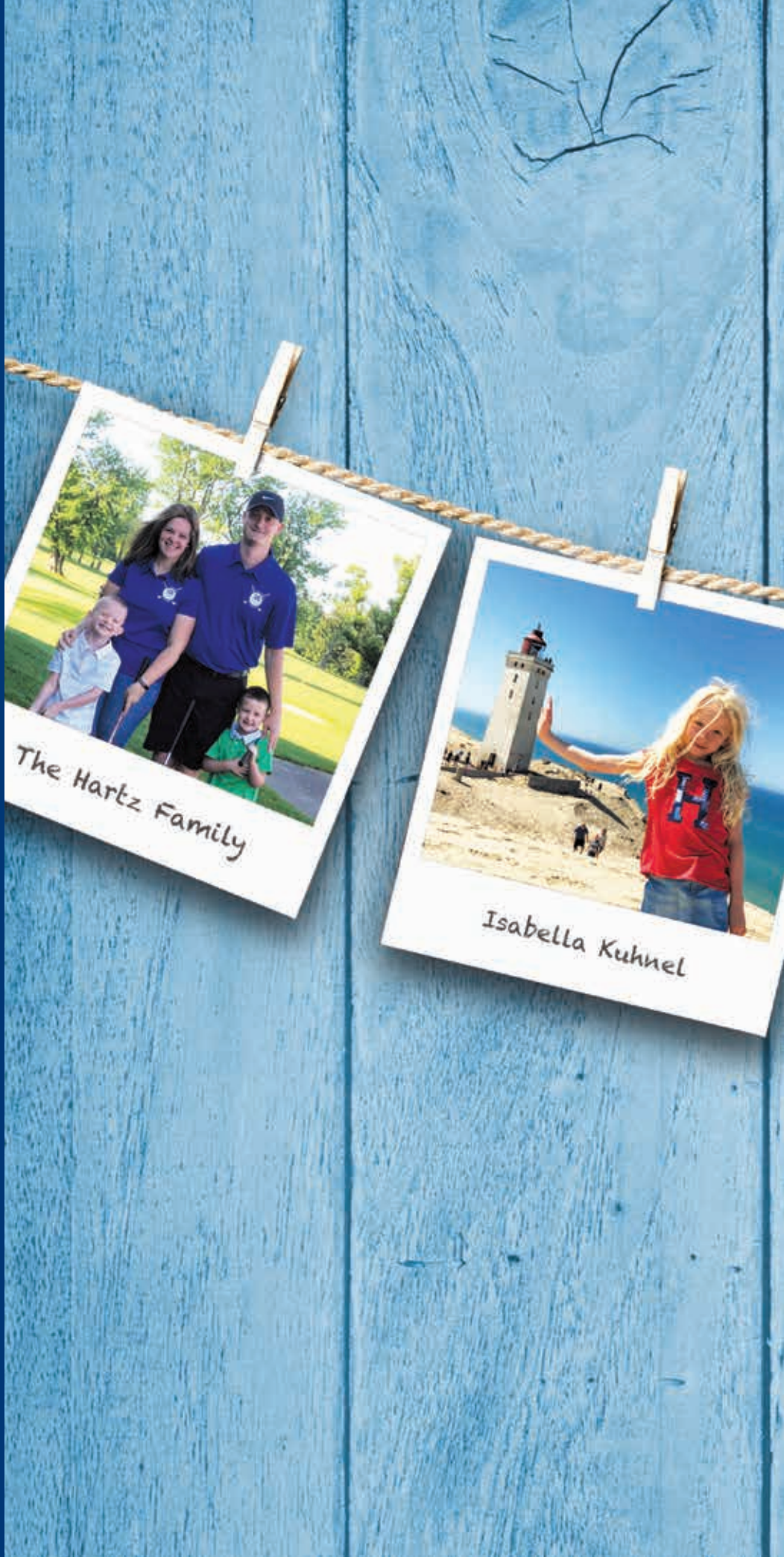
★★★★
CHARITY NAVIGATOR
Four Star Charity

WE'RE ON THIS JOURNEY TOGETHER

Thankful for our family, friends and community.

Grateful for the researchers who are dedicated to our community.

Hopeful that there will be a cure soon.



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