

cystinosis

magazine

For friends and supporters of the Cystinosis Research Foundation

Dream

COURAGEOUSLY



SUMMER 2017

DREAM COURAGEOUSLY ISSUE



Bryan and Alex are set to be married October 21, 2017

PAGE 42



DREAM COURAGEOUSLY

When hope is abundant, the cystinosis community is empowered to dream courageously—to dream of a future without cystinosis. Children can dream of being astronauts, parents can dream of meeting their grandchildren. With the research being done, and the driven families and friends fueling it, we know these dreams will one day come true.

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Dear Family & Friends



Summer is here, and with it comes a renewed sense of hope that we are one step closer to a cure. We watch our children with cystinosis dream courageously every day. Although their lives are filled with doctors' appointments, hospital stays, a myriad of different medications, they remain resilient and brave. Cystinosis does not define them; it makes them stronger. We have watched Natalie courageously battle cystinosis and are astonished by her ability to rise above the daily physical pain and choose to live her life to the fullest, dreaming of a future full of possibilities. Your love and support have given our children hope, and with hope, they bravely battle cystinosis.

We know you will enjoy this issue of the magazine. We have awe-inspiring stories of children and adults who are fighting back at cystinosis with their dreams for a better future. We have a special story of love in the cystinosis community, and we are eager to share the new grants funded last year. To help further acquaint you with the research being done and the people behind it, we have spotlighted three CRF-funded researchers who are working hard on our behalf to find better treatments and a cure for this disease. We are also honored to introduce two new cystinosis champions – people who have embraced our cause and helped make a difference in the lives of our families.

We revere the talent and dedication of those in the science and medical community who toil tirelessly for our cause. These people work every day to solve the mysteries of cystinosis. They think outside the box in their search for ground-breaking treatments. We have come so far and have built a strong foundation for innovative research which continues to expand with new findings and produce extraordinary results.

CRF's Commitment to Research

With your help, our science community is thriving. In 2003, when we issued our first research grant, we had a very specific goal: To find new treatments and a cure. Today, CRF is the largest provider of funds for cystinosis research in the world. We have awarded 151 multi-year research grants in 12 countries. Our researchers have published 67 articles in prestigious journals as a result of CRF funding. CRF-funded researchers work tirelessly on behalf of our community – they are committed to our children.

We receive dozens of research applications every year, which is a reminder that scientists and researchers from around the world want to solve the mysteries of this disease. We have invested your donations semi-annually to spur growth in our research and accelerate the research process. Our world-renowned Scientific Review Board reviews applications twice a year and after a thorough and comprehensive analysis, recommends studies for funding. We are indebted to our SRB members for their guidance and commitment to helping us fund the most talented researchers in the world.

Because cystinosis damages every cell in the body, our research portfolio is comprehensive, yet we have focused our research efforts on areas that cystinosis affects first and most severely – the kidneys and the eyes. We also target research that seeks answers on how to treat muscle wasting, neurological issues and endocrine complications of cystinosis.

We are able to study the complications of cystinosis by funding new grants twice a year. Last year, we established an accelerated research program that allows us to fund outside of our biannual schedule. This program will help us increase the pace of research and eliminate gaps in funding.

We are interested in finding new researchers and in funding understudied areas of cystinosis research, so we recently became a member of OnPar. OnPar is an organization in partnership with the National Institutes of Health (NIH) that offers funding groups like CRF an opportunity to support unfunded but highly rated NIH research proposals. Working with OnPar will enable us to fund new researchers and studies in areas of research that cross over with cystinosis, thereby allowing us to expand our research efforts.

We have created a synergistic science community and we have made significant progress. It is your commitment to CRF that has allowed us to fund studies year-round. We expect that very soon there will be more frequent discoveries and enhanced collaboration. Knowing that every minute of every day, somewhere in this world, a CRF-funded researcher is working on behalf of our children to save their lives is overwhelming.

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

A Banner Year for Research With Clinical Trials on the Horizon

We are pleased to announce that in 2016 we issued 17 new grants totaling over \$2.79 million in grant awards. The grant recipients for the fall 2016 are listed on page 74 along with a lay abstract of their studies. The areas of research involve new medications to treat cystinosis, understanding memory challenges in adults with cystinosis, learning more about kidney disease and cystinosis and studies focused on the pathogenesis of cystinosis. CRF researchers work collaboratively to learn more about cystinosis and how to treat it and cure it.

For years, we have supported bench research with the hope that one day that research would lead to clinical trials. We have hit our stride and are very close to realizing several new clinical trials. We are now in the position of applying what has been discovered in the lab to the bedside, to our children. It is directly because of your support of our mission that we are certain that clinical trials for novel treatments are on the horizon.

We are optimistic that within the next 18 months there will be clinical trials for corneal cystinosis, potential new drug therapies and stem cell and gene therapy. Your support has enabled us to translate promising scientific discoveries into potential new treatments that will lead to a cure!

STEM CELL AND GENE THERAPY

We have reported over the years about Dr. Stéphanie Cherqui's astonishing work using stem cells and gene therapy. We believe that bone marrow stem cells hold the promise of a cure for cystinosis. 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. We are grateful to the many volunteers with cystinosis who have donated their blood for this study so that Dr. Cherqui's group can determine the most efficient protocol to obtain corrected stem cells.

Dr. Cherqui, at the University of California, San Diego, continues to work with the Food and Drug Administration on the safety studies for this approach and we are hopeful that she will obtain FDA approval for a Phase I clinical trial for cystinosis this year.

We congratulate Dr. Cherqui for her recent grant of \$5.2 million from the California Institute of Regenerative Medicine. This CIRM grant ensures that Dr. Cherqui's work will be funded through the Phase I clinical trials. The grant is a great honor and is a tribute to her brilliance and CIRM's belief that Dr. Cherqui's work holds the promise for a cure for cystinosis. Dr. Cherqui has received a total of \$10.6 million

from other funding agencies. CRF's early commitment to her work provided the seed money necessary to attract other funding sources.

NOVEL TREATMENTS FOR CORNEAL CYSTINOSIS

We are close to realizing the success of two exceptional researchers, Ghanashyam Acharya, PhD, at Baylor College of Medicine; and Jennifer Simpson, MD, of the University of California, Irvine, who have been collaborating on a new treatment for corneal cystinosis. Dr. Acharya discovered a novel way to deliver cysteamine to the eye to treat 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 treatment but it is rigorous, painful for some and requires hourly dosing of medicated eye drops.

The nanowafer was designed by Dr. Acharya to deliver medication to the eye. The concept is to load the nanowafer with cysteamine, place it on the eye where the medication slowly releases, treating the eye for hours. CRF owns the license for the cysteamine-loaded wafer, which allows CRF to control the pace and direction of the research.

CRF's wholly-owned spinoff company, Corneal Cystinosis Inc., was formed to facilitate the continued development and clinical availability of the Cysteamine Nanowafer. The immediate goals of this effort are the formalization of manufacturing procedures, as well as the completion of pre-clinical testing required for initiation of a U.S. clinical study.

We are thrilled to report that we had a meeting with the FDA in April further advancing this project to a clinical phase.

CYSTINOSIS RESEARCH HELPS OTHERS

The work on cystinosis also brings new perspectives in the use of human stem cells in medicine as the findings on cystinosis allowed Dr. Cherqui to apply the same therapeutic strategy to other multi-organ degenerative disorders such as Friedreich's Ataxia, a neuro-muscular degenerative disease. Moreover, based on Dr. Cherqui's work, CRF-funded researcher Dr. Olivier Devuyst

in Switzerland recently reported that he could rescue the mouse model of Dent disease, an inherited kidney disorder, using the same stem-cell-mediated strategy.

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!

Committed to Finding Better Treatments and a Cure

The next few years of research are critical to our success in finding the cure. 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 hospital visits and blood draws, and without worries about life expectancy.

Your steadfast support, your commitment to research and your compassion and love for our community give us hope. Our children dream of a life free from cystinosis, a life they dare to live courageously with determination and grit.

We have made extraordinary progress and with your continued support and generosity we will soar to new heights and reach our goal of finding a cure for cystinosis.

Thank you for supporting cystinosis research, for standing by our side and for embracing our community. We are grateful for your partnership in our quest for the cure. We are blessed by your encouragement, your prayers and your unwavering belief that together we will find the cure.

With a grateful heart, we thank you,

Nancy and Jeff

DREAM COURAGE USLY



A Note from Natalie

Dear Family and Friends,

Since graduating from the University of Southern California last year with a Masters in Social Work, I have concentrated my efforts on acquiring more experience in the social services field as well as enjoying some time off to travel with my family and friends.

I interned at the Drew Child Development Corporation where I gained extensive legislative advocacy skills, which gave me insight into what I want to do with my career. I moved back to Orange County and now live with one of my best friends in Irvine, California.

I am excited to announce that in March I accepted my first full-time job. I am working at Taller San Jose Hope Builders, a nonprofit organization located in Santa Ana, California. Hope Builders advocates for young adults between the ages of 18 and 28 who have been through trauma at some point in their lives that has diminished their likelihood of employment success and sustainability. As the development manager at Hope Builders, I am responsible for corporate and individual donor cultivation, grant management, as well as strategy and performance management. I am thrilled to be starting my career in nonprofit management and cannot wait to see what the future holds for me.

As I start my career, I think about getting older and the fact that there is still no cure for cystinosis. I know the importance of taking my medications religiously and seeing my doctors annually. I also know the importance of my support system that will help me through whatever challenges may arise in the future.

The future is scary, but it will not stop me from pursuing my dreams and living my life as free of cystinosis as possible. I can and will always rely on my doctors, my friends, and my family for love and support. I am forever grateful to those in my life who have allowed me to have the courage to fight this disease and the faith that one day a cure will be found.

It will not stop me from pursuing my dreams and living my life as free of cystinosis as possible.

Our dedicated community is the reason we are so close to finding a cure. Because of committed parents, patients, and doctors, the battle is almost over. A life free of cystinosis is closer than ever before because of our awesome community.

I want to thank everyone in the cystinosis community for your unwavering generosity, kindness, and dedication to making my wish come true.

Love, Natalie Stack

*We CELEBRATE our CRF COMMUNITY
and are grateful every day for your support!*

CRF-FUNDED RESEARCH PROVIDES

HOPE!



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



WHAT IS CYSTINOSIS?

Cystinosis is a rare, inherited, metabolic disease that is characterized by the abnormal accumulation of the amino acid cystine in every cell in the body. Buildup of cystine in the cells eventually destroys all major organs of the body including the kidneys, liver, eyes, muscles, bone marrow, thyroid and brain.

Medication is available to control some of the symptoms of this terrible disease, but cystinosis remains incurable.

Cystinosis affects approximately 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 151 studies in 12 countries. **CRF has raised over \$39 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.

THANK YOU!

You have changed
THE COURSE
OF CYSTINOSIS

We are on the brink of new clinical trials and closer than ever to the cure. It is because of you that CRF has been able to fund extraordinary researchers across the globe.

IN 2016 ALONE, CRF FUNDED:

\$2.79 million
in research grants



17 scientific studies



SINCE 2003, CRF HAS ISSUED:

151 cystinosis
research grants
in 12 countries



1 step closer
to a cure



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.



CYSTINOSIS
COMMUNITY
CHAMPIONS

AMANDA McKIM
HIGH SCHOOL FRIEND

FRIENDS **FOREVER**



DURING A DARK SEASON, AMANDA McKIM KINDLES SOME CHRISTMAS MAGIC, FORGING AN UNBREAKABLE BOND WITH THE COE FAMILY

CHARLOTTE COE
Webster, New Hampshire



By Dennis Arp

It started with a play date. Megan Coe and Amanda McKim had been friends in high school, and now that they were reconnecting, they wanted their daughters to also enjoy some time together. So they met at a New Hampshire apple orchard, where their toddlers, Charlotte and Finley – both nearing a year old – quickly developed their own friendship. It was a great day, Coe remembers. But still, something was amiss.

“Our life with cystinosis was about to start,” Coe recalls. “Charlotte had not yet been diagnosed, but there were issues. We knew Charlotte was petite, and here she was with Finley, who was so much bigger. It was really eye-opening.”

Charlotte Coe’s diagnosis came shortly thereafter, and it hit everyone hard. The growing bond that Coe and McKim had rekindled became even more important.

“Amanda has always been so supportive,” Coe says.

McKim and her husband, Eric, started reading up on cystinosis and made a donation to the Cystinosis Research Foundation. They offered whatever help they could to Coe and her husband, Michael.

It didn’t feel like enough, but what more could they do?

Later, Coe shared a photo on Facebook that showed the number of medications Charlotte had to take. There was a basket full of syringes.

“So many syringes,” McKim remembers. “And all the other medications lined up.”

There were days when McKim would find herself weeping during her commute to work “just thinking about the struggles the Coes have and will continue to have until there is a cure for cystinosis.”

Then last year, as the holiday season approached, McKim got an idea. No matter the circumstances, no matter the struggles, she would ensure that Charlotte and the Coes enjoyed some Christmas magic.

“I know so many great people,” she thought.

So she and another friend from high school, Nicole Bryant, started compiling a wish list of gift ideas, which they included in a letter explaining the situation and their plan to surprise the Coes. That messaging went out in texts, emails, posts and face-to-face meetings.

“Boy did it take flight!” McKim says.

More than 50 people volunteered to help, some from as far away as Ohio and North Carolina. A neighbor McKim had never met came by with diapers and several outfits for Charlotte. One little girl got her family to redirect their usual gift exchange so that all 14 gifts could go to the Coes. Donations of money and needed items kept pouring in.



“We were able to anonymously fill their heating oil tank, pay a portion of their electric bill, get gift cards for gas, groceries and a home repair store, and so much more,” McKim says. “The value of all gifts reached \$5,000. I still get tears and goose bumps seeing that number.”

And the Coes didn’t know about any of it until Dec. 23. That’s when the McKims and others filled two cars with wrapped presents for delivery by Santa himself – Eric McKim, dressed as the jolly old elf.

“It was overwhelming and so touching,” Coe recalls of the moment when the surprise was revealed. “The cars pulled up packed with presents. We were outside in the snow, and I started crying, ‘This is all for us?’ We were just blown away.”

The presents for Charlotte were so thoughtful, and the gift cards eased family

burdens. But the most treasured delivery was a powerful message of hope.

“Just knowing we are loved by so many people in the community, that’s what means the most,” Coe says. “With this life, it can feel so lonely sometimes, because cystinosis is so rare. It’s hard for someone to truly understand, unless it’s another parent. How they coordinated everything, how they reached people who were eager to help our little family – that made us feel we weren’t alone. That was really the emotional part.”

These days, the Coes continue to have many bright lights in their lives. At age 2, Charlotte is walking and gaining weight. “All of her levels are in a good spot, so she is feeling as normal as she can, and that’s awesome,” Megan Coe says. “We hold onto these moments.”

Other cystinosis families the Coes met at CRF’s annual Day of Hope event are now treasured friends.

“We’re very fortunate to have this support and to know we’re not alone,” Megan Coe says. “And the work toward a cure is just amazing. All of these people who put in the time and effort for our kids, they are really like guardian angels for sure.”

“CRF’s Day of Hope was huge for us,” Coe adds. “In three days, we went from this feeling of devastation and heartbreak to hope and so much love. And it’s the same with Amanda. She showed us that people care – they really do care. This experience can be heavy and dark at times, but Amanda showed us light.”

Working together, Coe and McKim are now planning a fundraiser in their area to support the CRF and cystinosis research. It’s just one more indication that the Coes have a champion in their lives.

“I don’t even know how to put into words the depth of our friendship,” Megan Coe says. “Amanda goes above and beyond, and we’re just eternally grateful.”

For Charlotte and Finley, more play dates await.

“It’s a lifelong friendship,” Coe says. “We want our girls to be friends forever.”



CYSTINOSIS
COMMUNITY
CHAMPIONS

JENNY LEONARD

MANAGER OF CHOMP KITCHEN

WHERE LOVE SIZZLES ON THE GRILL



The "Lily Patty" features a house beef patty with cheddar cheese, spicy pickle aioli and nacho "Chompritos" (something like dust from Doritos), all inside a pretzel roll.

THANKS TO JENNY LEONARD, A BURGER JOINT BECOMES A HUB OF COMMUNITY SUPPORT FOR THE BEAUREGARD FAMILY, INCLUDING LILY, WHO GETS HER OWN NAMESAKE PATTY

LILY BEAUREGARD
Swansea, Massachusetts



By Dennis Arp

Jenny Leonard describes Chomp as “an amazing little burger joint in a tiny little town, where we love being a part of our community.” That love shows in a number of different ways but especially in Leonard’s appetite for turning regular customers into lifelong friends.

In fact, sometimes the love starts even before a customer is born.

“Kevin and Courtney used to come in for date nights when Courtney was pregnant,” Leonard says of the Beauregards, Chomp regulars. “So it’s fun to think that Lily developed on Chomp.”

As the manager at Chomp Kitchen and Drinks in Warren, Rhode Island, Leonard is the first smiling face restaurant visitors see. That smile is always more vivid when she sees 2-year-old Lily Beauregard coming in with her parents.

“Everyone turns and looks when Lily comes through the door,” Leonard says. “She’s so adorable, and she has a brightness about her. Now that she’s a bit older, her personality really comes out. Like her mom and dad, she’s a caring person – her eyes and her smile give that away.”

But not every encounter prompts expressions of joy. On one visit, the Beauregards shared the news that Lily had been diagnosed with cystinosis.

“You could see that it was difficult for them to talk about it,” Leonard recalls. “By the end of the conversation, I was in tears.”

Leonard wanted to help, so Kevin Beauregard came up with an idea. He had noticed that on occasion Chomp would designate a special “charity burger,” with proceeds from its sale going to aid a particular cause. Would the restaurant consider cooking something up to support the Cystinosis Research Foundation?

“It’s not something I had to think twice about,” Leonard says. “I didn’t even have to ask my boss (Sam Glynn). I knew Sam would be ready to move forward.”

The next step for Leonard was to learn all about cystinosis. She discovered that in addition to the debilitating effects of the condition, patients often crave salty and cheesy foods.

“My chef is talented, and I knew he would take that information and run with it,” Leonard says. “So he started working on the ‘Lily Patty.’ The next thing you know, he comes out with a new burger and says, ‘What do you think of this?’ I said, ‘I think you’re a genius.’”

Chef Mike Oozoonian’s special creation featured a house beef patty with cheddar cheese, spicy pickle aioli and nacho “Chompritos” (something like dust from Doritos), all inside a pretzel roll. “Mmm,” Leonard said. Hmm, the Beauregards thought.

“We looked at each other like, I don’t know if that sounds so good,” Courtney Beauregard remembers. “Then we had one. The chef is amazing. He melded everything together in this perfect burger.”

Leonard took things from there. She picked a month for the Lily Patty to be on the menu and then wrote up a description, not only of the burger but of the cause it supported. She went the extra mile of educating the wait staff about cystinosis and how it afflicts children, including one of their neighbors.

“She made sure there were pamphlets and that everyone there learned so they could educate others,” Courtney says. “That meant a lot. There were a lot of people who otherwise might have ordered it in passing, but now they cared.”

The Lily Patty quickly became Chomp’s most popular burger throughout the

month, but even so, “I didn’t feel like it was enough,” Leonard says. “I wanted to do something bigger. So I told my boss that we should do a whole night for Lily. We did specials and really promoted it, with 20 percent of each burger sale going to cystinosis research. It was one of our busiest nights. Whole families came – all of our regulars came. It was a great way to spread awareness.”

All told, the Lily Patty raised \$1,200 for CRF and cystinosis research. But the effort also provided something else. It showed the Beauregards that their community cares deeply about Lily and the cause of cystinosis research.

“It excited me so that I want to do more fundraisers,” Courtney says.

A sense of community now pervades the Beauregards’ lives. Through CRF, social media and Lily’s physician, Dr. Deborah Stein at Boston Children’s Hospital, they’ve developed a support network that includes other cystinosis families. That community is helping Lily to “blossom,” Courtney says, despite the many challenges of her condition.

“She’s growing now, and even when she’s sick she’s usually in a good mood,” Courtney notes. “We’re extremely fortunate.”

It helps that close to home, they have a true champion in Jenny Leonard.

“We check in with her a lot,” Courtney says. “It’s not a customer relationship; it’s a deep friendship.” Likewise for Leonard, Lily and her family are much more than burger buddies.

“I absolutely adore Lily and love them as a family,” she says. “I tell them all the time that I’m so thankful and honored to know them, and I’m eager to keep going on their journey with them for as long as I can.”



CHOMP
KITCHEN AND DRINKS

Better Sleep, Better Memory?



Doris Trauner, MD

PROFESSOR, DEPARTMENT OF NEUROSCIENCES
UNIVERSITY OF CALIFORNIA, SAN DIEGO

Dr. Doris Trauner has a deep connection to the brain health of young patients. Now she begins a study aimed at improving the lives of adults with cystinosis.

As a pediatric neurologist, Dr. Doris Trauner explores the cognitive effects of conditions that alter brain development early in life. She has been drawn to cystinosis since she wrote her first paper on the subject in 1988, but she's also the first to admit there's so much more to learn, especially about adults with cystinosis. That's why opportunities to perform new research are so important.

With grant support from the Cystinosis Research Foundation (CRF), Dr. Trauner is preparing to study the possible connection between sleep disturbance and memory problems in adult cystinosis patients. As with so much of her work, the results could have a direct effect on quality of life.

"If patients continually have problems getting to sleep and staying asleep, it can affect attention and memory, which are problems we're seeing anecdotally," said Dr. Trauner, distinguished professor in the departments of neurosciences and pediatrics at the University of California, San Diego School of Medicine. "The idea of the study is to look at these variables and see if sleep problems correlate with memory issues."

If the connection holds, treatments for sleep disturbance might be all that's needed to also bolster memory and concentration.

The link between cystinosis and cognitive issues is already well established, Dr. Trauner says. Virtually all lysosomal storage diseases have adverse effects on brain function, and since the late 1980s doctors treating young cystinosis patients have seen evidence of problems with fine motor skills and coordination, gross motor delays, low muscle tone, and on occasion even tremors and seizures.

"A specific learning profile has developed," the doctor said. "IQ and language functions tend to be normal, but sometimes there are

problems with visual spatial processing and memory. We've done MRI brain imaging and found that the part of the brain responsible for special processing develops differently. My theory is that the gene for cystinosis also affects brain development."

The cognitive differences can cause children to struggle in school, particularly with arithmetic and spelling. Later, there can be problems with tasks like reading maps and finding a car in a parking lot, or getting around in any unfamiliar place, Dr. Trauner said.

"A small percentage of patients have increased pressure in the brain," she added. "As they get older into adult life, there are neuromuscular issues and problems with muscle weakness."

Cystine accumulation over time breaks down all muscle cells. In addition to problems such as with grip strength, dexterity and endurance, complications can become life-threatening when they grow to include functions as elemental as swallowing. Deterioration of these muscles can even trigger aspiration of food and saliva into the lungs.

"As people live longer with cystinosis and in general are healthier thanks to advances in treatment, some of these other problems are surfacing, and neurological problems are among the more common," Dr. Trauner said.

"Now we are encountering more widespread memory problems – forgetting what you did yesterday, not being able to remember an assignment, that sort of thing.

"If there is an association between sleep and memory, depending on the types of problems, we might be able to suggest a specific treatment," she added. "But first we have to differentiate, because they all have different treatments."

For the year-long CRF-funded research project, Dr. Trauner is hoping to recruit 15 or more adult cystinosis patients to come to San Diego and participate in an overnight sleep study. Enrollees will take part in three

to four hours of cognitive testing, including a baseline IQ testing and several kinds of memory and neuropsychology tests.

Then patients will go to a sleep lab, where devices will measure oxygen in the blood, respiratory rate and depth of breathing. Electrodes on legs and arms will help determine how much participants move during sleep, while those on the scalp monitor brain activity.

A sleep specialist is part of the team to maximize analysis of the data gathered during the night.

The study is expected to shed light on a topic of growing concern for cystinosis patients. But no matter what the study finds, Dr. Trauner is eager to help grow the overall body of knowledge about a disorder that affects patients and families who are close to her heart.

AS PEOPLE LIVE LONGER WITH CYSTINOSIS AND IN GENERAL ARE HEALTHIER THANKS TO ADVANCES IN TREATMENT, SOME OF THESE OTHER PROBLEMS ARE SURFACING, AND NEUROLOGICAL PROBLEMS ARE AMONG THE MORE COMMON.

"When I first became aware of cystinosis, most patients were not living into adult life," she said. "So we've made huge strides, which is very exciting. And that wouldn't have happened without the CRF, which raises more money than the National Institutes of Health (NIH) pays to all research on cystinosis.

"When I hear from families with specific questions, I think about all the specialists and how these days we have better answers to give," she added.

The best news is that as Dr. Trauner moves forward, there may be more answers to share and more treatments to recommend.

Muscle and Bone



Mary Leonard, MD, MSCE

PROFESSOR AND CHAIR, DEPARTMENT OF PEDIATRICS
STANFORD UNIVERSITY

A largest-ever study sheds light on a critical connection and the importance of weight-bearing exercise.

For the first time, a comprehensive research project has used high-tech scanning equipment to explore the ways cystinosis affects bone and muscle development. And right from the start, it became clear that such a study was long overdue.

“Strikingly little is known about bone health in cystinosis patients,” says physician and researcher Dr. Mary Leonard, Chair of the Department of Pediatrics at Stanford University. “Medical literature includes a handful of studies, some with as few as two or three patients. The biggest included nine children. Early on, we realized that we don’t even know what normal is – how thick should bones be in a 9-year-old?”

Thanks to funding from the Cystinosis Research Foundation (CRF) and support from Dr. Paul Grimm and the committed cystinosis community at Stanford, Dr. Leonard’s project was able to enroll 38 patients – more than four times as many as in the largest previous study. The results are still being evaluated, but already they are offering insights that could lead to multi-center trials of specific interventions.

“We will use this information to help physicians understand risk factors for poor bone development and prepare for clinical trials to target these risk factors,” Dr. Leonard says.

In broad strokes, the research shows that bone mineral density is quite low in cystinosis patients, with more than half the participants ranking below the 10th percentile for their age. One driver is diminished kidney function, Dr. Leonard says, but there’s also new evidence of a connection between muscle weakness and bone health.

“For the first time we really have an appreciation for how poor patients’ muscle strength is,” Dr. Leonard says. “Muscle is so important for bone development. The force muscles exert on bones triggers changes in bone metabolism to make them stronger.”

The muscle weakness was greater than researchers expected.

“It makes us wonder if we can improve bone health by working on muscle strength,” Dr. Leonard says. “It’s just observations, but the two do seem to go hand in hand. With all of our kids whose bone density is below normal, their muscle strength is very much below normal.”

Weight-bearing exercises are the most immediate ways to improve muscle mass, Dr. Leonard said. Because children with chronic kidney

disease in general are at a higher risk of heart disease, Dr. Leonard recommends regular physical activity anyway. “Parents ask a lot about swimming, and if that’s what your child wants to do, great. Any exercise is important,” she says. “But it isn’t as good for bone health as weight-bearing physical activity.”

That leaves room for lots of athletic choices, although Dr. Leonard adds a key proviso: Be careful with fragile bones.

EARLY ON, WE REALIZED THAT WE DON'T EVEN KNOW WHAT NORMAL IS – HOW THICK SHOULD BONES BE IN A 9-YEAR-OLD?

“You probably don’t want them out there on the ice swinging sticks playing hockey, or playing football,” she says. “I’d avoid contact sports.”

As for the question of supplements, Dr. Leonard says that physicians treating cystinosis patients are already working hard to keep phosphorous levels in the healthy range. She’s waiting for more findings from some of the study’s more sophisticated blood tests, but she doesn’t expect that they will change her recommendation of no new supplements.

One of the factors that distinguishes this research project is its use of advanced tools, including DXA bone density scans. Such scans are an enhanced form of X-ray that gives a deeper and more detailed look at bone and muscle health.

The researchers plan to publish on their findings. Use of the scans and what they reveal will be the source of one paper, while insights related to the fine microarchitecture of bones will be at the heart of the other published report.

One thing she does know: The research project would not have been possible without CRF funding as well as the cystinosis community’s support for patient participants.

“Never in my career have I seen and worked with a foundation that was able to mobilize around research and support the way this group does,” Dr. Leonard says. “This group of families came together in such a supportive, productive and collegial way. So it’s not just the financial support, which is vital – it’s the overall support, which is impressive.”

Lower Peak, Better Price



Patrice Rioux, MD, PhD
CHIEF MEDICAL OFFICER
THIOGENESIS THERAPEUTICS, INC.



Vincent Stanton, MD
CHIEF SCIENTIFIC OFFICER
THIOGENESIS THERAPEUTICS, INC.

What if delayed-release cysteamine had a steadier delivery and cost a lot less? Drs. Rioux and Stanton are working on that.

During his two decades in the United States as a pioneering researcher and pharmaceutical industry executive, Patrice Rioux, MD, PhD, has touched countless lives with his tireless work. Patient need has always topped his list of priorities, but there was a time when personal ambition also shaped his definition of success. Not anymore.

“My partner (Dr. Vincent Stanton) and I are not trying to make our careers – we’re two old guys,” Dr. Rioux says with a laugh. “We just want to do something useful for patients. Maybe if I were 25 years old again, I might have a different motivation. But now I just want better lives for patients and their families.”

Aided by a grant from the Cystinosis Research Foundation (CRF), Dr. Rioux is getting that chance. He and Dr. Stanton are co-principal investigators on a new project that will test the properties of several precursors to cysteamine, the predominant drug used to treat cystinosis.

It turns out that cysteamine is produced naturally in the body, although at very low levels. The goal of their research is to develop a new medication using some of the natural substances that can be degraded to cysteamine, which should lead to a kind of natural delayed-release cysteamine, which combats the buildup of cystine crystals in cells while extending the time between dosages. Non-natural precursors will also be tested.

Dr. Rioux was chief medical officer at Raptor Pharmaceuticals, Inc., where, with research funding support from the CRF, he led the U.S. and European clinical trials and secured regulatory approval for the delayed-release version of cysteamine. The resulting product has the brand name Procsybi.®

“I was very pleased to work on that project with Drs. Jerry Schneider and Ranjan Dohil. It was great to be able to improve the lives of cystinosis patients,” Dr. Rioux says, adding “But I wasn’t happy with the way the drug was eventually priced, and that prompted my departure.”

The delayed-release formulation allows for twice-a-day dosing and so gives patients the chance to sleep through the night, making compliance that much easier and lives a whole lot better.

But there are still opportunities for improvements, Dr. Rioux says. A key goal is to reduce the drug’s peak concentration, which usually occurs within the first two hours and is associated with many of the familiar side effects of cysteamine.

WE WANT A MEDICATION THAT IMPROVES PATIENTS’ LIVES AND THAT FAMILIES CAN AFFORD.

“Right now, for the drug to have enough exposure, there has to be a huge peak,” he says. “Then as soon as the level of cysteamine drops, it loses effectiveness.”

Thus, by producing a more even drug concentration Dr. Rioux hopes to reduce the drug’s side effects, including stomach distress and other pressures on the gastrointestinal tract as well as the bad breath and body odor that often accompany use.

By using precursors for their research, “instead of just changing the presentation of the capsule we’ll actually be using a different compound that turns into cysteamine,” Dr. Rioux says. “We think we can achieve absorption with a lower peak and fewer side effects while still seeing a long exposure.”

Because the resulting medication will be effectively a new compound, Drs. Rioux

and Stanton, as principals at Thiogenesis Therapeutics, Inc., are expecting to greatly control costs. That will translate to substantial savings for families.

“That’s the reason just the two of us are working on this,” Dr. Rioux says. “We would like to keep at least 50 percent control so we can avoid capital coming in saying we should sell at a higher price.”

When the time arrives for clinical trials and then for navigating the complicated process of regulatory approvals, significant capital still is a necessity, Dr. Rioux notes.

“But when I see crazy prices (for other medications, especially in the rare disease space), it becomes clear what we are determined to avoid,” he adds.

To maximize their chances of success, the researchers plan to test two or three cysteamine precursors to find which provides the longest exposure with the lowest peak that is still therapeutically effective. Because the biochemistry and physiology of cysteamine metabolism have been studied extensively, the project should move quickly from testing with rats to mini pigs (whose GI system is closer to that of people) to human patients, Dr. Rioux says.

“If it goes well, we should have patients in clinical trials in 2018,” he notes. “We hope to be approved three or four years from now. We’re really just starting, so you never know, but we’re optimistic.”

The vast amount of industry experience represented by Drs. Rioux and Stanton, especially with regard to cystinosis and cysteamine, should also expedite the process.

“The great thing about collaborating with the CRF is that we’re all working for the benefit of families,” Dr. Rioux says. “We want a medication that improves patients’ lives and that families can afford.”

“Some drugs are necessarily more expensive,” he adds. “This one shouldn’t be.”



Canada Unites with CRF To Find A Cure

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.

Canadians can send donations directly to CRF or they can contribute to CRF through the Canadian Aqueduct Foundation. Through the Aqueduct Foundation, Canadians have created an efficient and effective fundraising process which allows them to fundraise, ensure their donors receive a charitable tax receipt and fund CRF Scientific Review Board-approved research studies.

The Cystinosis Awareness and Research Effort (CARE) and the Liv-A-Little Foundation have a strong working relationship with CRF. Jody Strauss (CARE) and Erin Little (Liv-A-Little) are CRF Board of Trustee members.

CRF is the largest fund provider of cystinosis research in the world. CRF has created a world-renowned scientific review board composed of leading cystinosis scientists and experts who review and recommend research applications for funding. CRF is committed to funding the most promising and innovative research. CRF is thankful for the Canadian families who have joined CRF in an effort to find better treatments and a cure.

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

- Valeri 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 **\$350,512.75**.

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

Robert Mak, MD
University of California, San Diego
 “Leptin Signaling in Infantile Nephropathic Cystinosis”
 \$74,989.75

Liam Feng, PhD
Stanford University, California
 “Molecular Mechanisms of Cystinosis”
 \$37,500

Alan Davidson, PhD
University of Auckland
 “Kidney Organoids: A New Model to Study Cystinosis” \$36,094
 Funded by Liv-A-Little Foundation

If you would like to learn more about how to fundraise in Canada, please contact Erin Little (ce.little@bmts.com) or Jody Strauss at (jody.strauss@yahoo.ca)





day of
H O P E



2017

CYSTINOSIS RESEARCH FOUNDATION
FAMILY CONFERENCE



MEDICAL ADVANCES

HIGHLIGHT

CRF's ANNUAL

DAY OF HOPE

The 2017 Day of Hope Family Conference was a tremendous success. Fifty-six families from around the world gathered in Newport Beach, California to learn, share, laugh and celebrate for three inspiring days with fellow members of the cystinosis community.

The hopes and dreams of attendees were shared at the conference in simple declarations hand-printed on colorful construction paper circles of pink, purple, blue and green – heartfelt wishes that soared buoyantly to the left of the lectern where researchers presented their latest findings about the disease:

Thankful for being healthy today.

I'm grateful for all the friends I met here.

My hope is that there will soon be a cure.

These are the familiar words of those whose lives have been touched by cystinosis, but they are also reflective of the hope that comes with new friendships, a growing network of support, and groundbreaking advances in CRF-funded research on the pathology and treatment of the disease.

day of HOPE

Recent discoveries are making it more possible than ever for those with cystinosis to feel healthy. The sense of community established through the Cystinosis Research Foundation has resulted in many lifelong friendships and special bonds, and a greater understanding of the causes of the disease has researchers closer than ever to finding a cure.

“When we started, we would fund two, four or six projects a year,” CRF President Nancy Stack told a packed ballroom at the Island Hotel. “Now, we are funding research worldwide and issuing new grants twice a year. Last year we raised over \$4.9 million which will allow us to continue to fund important and ground-breaking research.”

Since 2003, CRF has issued 151 multi-year research grants in 12 countries. Its researchers have published 67 articles in prestigious medical journals. The foundation has established the Cure Cystinosis International Registry, sponsored a biennial CRF International Research symposium of scientists throughout the world, and is poised to get clearance from the Federal Drug Administration for a new clinical trial very soon, Stack said.

Some highlights coming from this year’s research include:

Advances in delayed-release cysteamine treatments

The first patient pilot study for allogeneic stem cell and gene therapy

Potential new treatments for corneal cystinosis

Developments in a transdermal patch for cysteamine

Ground-breaking bone and muscle studies

Speakers throughout the three inspiring days included medical doctors and researchers who presented encouraging findings on the latest treatments, advancements and insights related to the disease.





Among the many speakers, Stéphanie Cherqui, PhD, of the University of California, San Diego, presented exciting news about an upcoming clinical trial on stem cell gene therapy for cystinosis.

Sergio Catz, PhD, of The Scripps Research Institute, delivered information on new insights into the trafficking of the lysosomal autophagy receptor, which may lead to new therapies for the disease.

Robert Mak, MD, also of UCSD, described new strategies for treating inflammation and the inflammasome in cystinosis; and Jennifer Simpson, MD, of the University of California, Irvine, and Ron Kurtz, MD, of Corneal Cystinosis, Inc., described the ocular manifestations and treatments of cystinosis in the eyes.

“There’s a lot of things in the pipeline and a lot of reasons to have hope,” said Stephen Jenkins, MD, a member of the CRF Board of Trustees, who spoke on the effects of cystinosis on all cells in the body and provided an overview of recent medical advances. As a parent of two sons diagnosed with the disease, he urged everyone to stay informed.

“We all have a responsibility to learn everything we can for our families,” he said.

Paul Grimm, MD, of Stanford University, provided tips to families on managing the symptoms of Fanconi Syndrome and the problems that can arise when medications and supplements interact negatively with each other.

The overwhelming schedule of caring for a child with cystinosis can present a challenging juggling act that is unique to each individual.

“All cystinosis patients are not the same,” Grimm said, addressing the many parents who traveled from throughout North America and beyond to attend the conference.

“Don’t feel guilty if you feel you’re doing something wrong, and don’t compare your child to another one. Above all, maintain good communication with your doctor and stay on top of the latest findings and developments about the disease. We’re all in this together. We’re all one united community.”

More complete information about CRF-funded research and discoveries can be found on pages 70 and 74.



NATALIE'S *Wish* CELEBRATION

one wish. one night. one step closer.

A Night of Extraordinary Generosity and Hope

Hope filled the room on Saturday, April 1, as 475 guests from around the world came together for the cystinosis community's largest fundraiser, the Natalie's Wish Celebration at The Island Hotel in Newport Beach, California. The event was a record-breaking success, raising \$3.6 million for cystinosis research.

Guests entered a beautiful and captivating ballroom with hues of purple and blue decorated with a message that captured the inspiring spirit of the evening: One Wish. One Night. One Step Closer.

Attendees traveled from across the globe including 56 cystinosis families from Sweden, Canada, France, Ireland, Australia and Norway to celebrate the milestones we have reached, the discoveries we have funded and to focus on the incredible work still ahead. The gala was the finale to the three-day Day of Hope family conference. Each year, families who have raised funds for CRF in their own communities proudly parade on stage to present their checks. From bake sales to golf tournaments, this community is unstoppable.

The successful evening was highlighted by a special performance from Us The Duo, whose latest album, "Just Love,"

recently hit No.4 on the iTunes Pop Chart. Another special part of the evening was the energetic live auction which featured items including fine dining, luxurious getaways and extraordinary wine collections, raising more than \$291,000.

The night was filled with moving stories from cystinosis community members including Natalie Stack, who shared an update on where she is now, as a newly employed young professional at a nonprofit in Santa Ana, California. Natalie beamed as she thanked members of the community for their continuous support that has led her to where she is today. Her mother, Nancy Stack, then shared incredible milestones the community has reached and scientific discoveries the community has funded. She thanked guests for helping to make Natalie's wish and the wish of all those with cystinosis come true.

Later in the evening, Ashton Jenkins, mother of Sam (7) and Lars (4), both with cystinosis, shared her family's journey through treatment and how strong and brave Sam and Lars truly are for battling cystinosis every day. She spoke of the ups and downs, and how the dedication and perseverance of CRF fills her with hope that a cure is within reach.





A Mother's Message of Courage:

A few weeks ago, my 4-year-old, Lars, who has cystinosis, got home from preschool and asked me, "Mom, did you know that there are kids in my class who don't swallow pills? Ever?!" With a huge smile on his face he asked me when he could stop taking medicine. It hurt my heart to tell him that the 16 big blue pills he swallows every day are essential to keeping his body healthy.

Lars has a 7-year-old brother who has charted the cystinosis path before him. Samuel was diagnosed with cystinosis when he was just a year old. Lars was only 2 when he watched his cool older brother swallow more than 20 pills each morning before school. Lars was determined to be like his brother and learn to swallow pills. He started practicing with mini M&M's and never gave up. Before long he was swallowing five Cystagon® pills every six hours.

Some days the boys make it look easy but it never really is. My husband and I make up crazy stories and pretend pills are dinosaur eggs and Lars is the T-rex coming to eat them. If that doesn't work, we'll pretend that the blue pills contain super powers and the white pills are alligator teeth. Other times I will sit with the boys for 30 minutes coaching, encouraging, begging and sometimes bribing them to take their medicine on schedule. Some days are

especially hard because their stomachs are aching, they have already thrown up once or twice, and they are just plain tired of swallowing pills.

I wish that the pills they take each day could make them feel better immediately. Then they would more easily understand how important the medicine is to keep them healthy. Unfortunately, every medication has side effects, and more often than not the pills make the boys feel worse.

Take potassium, for example. Because children with cystinosis lose so much potassium in their urine, we have to give Sam enormous doses to keep his potassium levels in the normal range. Each month I pick up a grocery bag filled with eight large bottles of potassium chloride from the Primary Children's Hospital pharmacy. More than once the pharmacy technician has made me wait while a concerned pharmacist calls our nephrologist to confirm that Sam really takes the absurd amount of potassium prescribed for him.

The potassium supplements upset Sam's stomach. He has thrown up many times at school but tells me it's OK because it's almost always below the playground where nobody can see. I used



ASHTON JENKINS' SPEECH AT
NATALIE'S WISH CELEBRATION

to assure him that kids don't remember those things until I introduced him to our new neighbor who said, "Yeah, I know him. He's the kid who threw up in the lunchroom."

When Sam was first diagnosed almost seven years ago, some people told us to never use the word "cure," because it was unrealistic and would spread false hope. After attending our first Cystinosis Research Foundation conference, however, we were inspired by the dedication and perseverance of the Stack family and Dr. Stéphanie Cherqui, and we left filled with hope that we really can beat this disease.

We are so grateful to Nancy and Jeff Stack, and to all of you, who generously support this cause. It is your encouragement and love that I remember in moments when I feel like this disease is a burden too heavy to bear. I hope that one day I will be able to look at my son Lars and tell him he doesn't have to take pills anymore. Thank you.

Music Ignites Hope:

This year's 2017 Natalie's Wish Celebration featured an amazing performance by acoustic sensation, Us The Duo. The husband-and-wife musical team, Michael and Carissa Alvarado, played many songs including hits from their new album "Just Love" which recently hit No. 4 on the iTunes Pop Chart. Us the Duo recently wrapped up an international tour with Pentatonix, a five-member American a cappella sensation.

Before their performance, the pair met with cystinosis families and patients and learned about their stories.

"This community is filled with warriors. Each and every cystinosis child, young adult and family member we met exudes a contagious sense of optimism and strength," shared Carissa. "My heart goes out to these families. One of our songs, "No Matter Where You Are," resonated with us as a perfect song to describe the love and support woven into the cystinosis community."

*I will stand by you
Even when we fall
I will be the rock, that holds you up
and lifts you high so you stand tall*

*I won't let you go
No one can take your place
a couple fights & lonely nights
Don't make it right to let it go to waste*

*I won't let you fall
I won't let you go
No matter where you are
No matter where you are, I'll be there*

*I will hold on to
Everything we've got
A quitter, a regretter, and forgetter is
Everything I'm not*

*I'll take care of you
and love you just because
You and I are better than forever
Nothing can stop us*

I'll be there I'll be there



"We are lucky enough to play music around the world and it is amazing how the power of a guitar or a voice is universal. It really brings people together," shared Michael. "We craft songs that can help change people for the better and provide them with hope through tough times."

"Seeing the courage and love in this room tonight has inspired us to go home and write more," added Michael. "You can see the fight in each and every person in this community, and we are honored to have been a part of Natalie's Wish."

THANK YOU!

You have changed

THE COURSE OF CYSTINOSIS.



The Cystinosis Research Foundation is eternally grateful to all its 2017 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

Traci and Tom Gendron, *in honor of Tanner Edwards*

Nancy and Jeff Stack

\$450,000 – \$499,999

Anonymous

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Liv-A-Little Foundation, *in honor of Olivia Little*

Jenna & Patrick's Foundation of Hope, *in honor of Jenna and Patrick Partington*

David A. Pyle Family

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Tina's Hope for a Cure, *in honor of Tina Flerchinger*

Laura Khouri and Michael K. Hayde

Susan and Tom Moran

24 Hours for Hank, *in honor of Henry Sturgis*

\$50,000 – \$99,000

Hearts for Hadley, *in honor of Hadley Alexander*

Curl for a Cure, *in honor of Lola Long*

Chris and Bill Thormahlen

Sue Werner and John Hagestad

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Fore Fathers, *in honor of Andrew Cunningham*

Seth's Circle of Hope, *in honor of Seth de Bruyn*

Hope for Holt, *in honor of Holt Grier*

Lots of Love for Landon, *in honor of Landon Hartz*

Nelda Arnold and W. Dean Henry

Peter K. Kompaniez

Jill and John Manly

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Mary and Peter Rooney

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The Emerson Family, *in honor of Brooke Emerson*

Music for Mary, *in honor of Mary Head*

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 Betty and David Shank, *in honor of*
Rebecca Shank
 Lynda and Ken Stack
 Kathryn A. Stack
 Tammy and Rodney Stephenson,
in honor of Son Gabe's 21st Birthday
 Dr. Doris Trauner and Richard Stanford
 Ann and Marc Winthrop
 Joan and Jim Ziegler

\$250 – \$499

Janine and Dennis Arp
 Hinda and Hal Beral
 Marlene Bitanga
 Cylilia Boren and Jerry Tadlock, *in honor*
of Jordan Darling
 Kristina Broadbelt
 Jennifer Doyle
 Daniel Dunn
 Cyndee and Jon Ely
 Bruno Gagnon
 Aimee and Bob Klaewtanong
 Steve Longo
 Jan and Scott McPherson
 Marylyn Milburn
 Eric Mosbrooker
 Kelly and Eric Olsen

Jennifer and Jamie Peachman, *in honor of*
Morgan Peachman
 Karen and Frank Ritchie, *in honor of*
Frank and Zack Ritchie
 Mari and Col. Ray Snyder (Ret)
 Rhonda and Craig Tudor

\$100 – \$249

Marcu and Ben Alexander, *in honor of*
Hadley Alexander
 Lynn and Art Barrett
 Lora Carpenter
 Jenny and Dr. Sergio Catz
 Hollie and Kerry Christopher
 Jeff Cole
 Joan and Robert Curtin
 Don and Karen Cunningham, *in honor of*
Andrew Cunningham
 Marthe Drolet, *in honor of*
Marilynn Lepack
 Sue and Peter Dunsdon
 Timothy J. Gleason
 Lisa Dorsey Glass
 Mary and Jim Harrington
 Pamela and Jeffrey Howard
 Jillian and Robb Hunt
 Nicole Inal
 Erica and Joshua Johnson
 Chris and Roger Jolicoeur
 Barbara and Jeff Kulyk, *in honor of*
Jordan Janz
 Leslee and Lewis Levey
 Sue and Andy Maguire, *in honor of Ciara*
and Sinead Maguire
 Sandy and Jerry Martin
 Shaun Nichols
 Anne Marie O'Dowd, *in honor of*
Luke O'Dowd
 Gail and Marty O'Hea
 Kaitlin Geuss and Ed Piddington
 Vicki and John Rezzo
 Barbara and Trevor Rodgers
 Andrea Shrewsbury, *in honor of*
Kennedy Hughes
 Lauren and Robert Silvernail
 Linda L. Stack
 Bethann C. Sullivan
 Jennifer Tetley

Gifts in Kind

Colgin Cellars, Ann Colgin and
 Joe Wender
 Richard Eddy
 Chef Alan Greeley and
 Chef Alessandro Pirozzi
 Harlan Estate, Bill Harlan
 Laura Khouri and Michael K. Hayde
 Lugano Diamonds, Idit and Moti Ferder
 Meadowood Napa Valley
 Napa Valley Reserve, Carol and
 Philip Norfleet
 Neiman Marcus, Fashion Island, Leyla
 Vokhshoori, V.P., General Manager
 Chef Tony Nguyen
 Christyne and Bob Olson
 Orange County Business Journal
 Promontory Estate, Charlie Williams
 Nancy and Jeff Stack
 Tejon Ranch Company



cystinosisresearch.org 949 223 7610

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*“Help Lily Blossom”
will officially be
standing strong with
so many of you who
dream courageously.*

Joining the Ranks of Those who *Dream*



By Kevin and Courtney Beaugard, Lily's parents
SWANSEA, MASSACHUSETTS

Our perfect little girl arrived in April of 2015. The joy we felt was more than we could have ever expected.

When Lily was around 3 months old, she started spitting up more than normal, especially first thing in the morning. After months of trying different formulas and medication with no resolution, we saw a gastroenterologist. Months passed and we made three more trips to the GI specialist only to be told that we were just nervous new parents and our child was perfectly healthy.

We arrived at her 12-month check-up armed with a list of different specialists we wanted Lily to see. We knew in our gut that something was wrong with our girl, and we would fight until we found out what it was. At that very appointment, Lily was diagnosed with failure to thrive. We left with an appointment to see a new GI specialist and neurologist out of Boston Children's Hospital. After a barium swallow test, we received a call a few days later. We were told they wanted Lily to be admitted to the hospital because they thought she had rickets. After just three days, multiple lab tests and visits with many different doctors, we were given the diagnosis of cystinosis. As relieved as we were to have a diagnosis, we were not prepared for the words, "Your daughter has an ultra-rare genetic disorder and there is no cure." After speaking to the nephrologist at Boston Children's for what seemed like hours, we knew we were in the right hands and that Lily would have excellent treatment. This was the beginning of our new chapter.

We are now nine months into this new journey and Lily continues to impress and inspire us on a daily basis.

Lily is a strong, sassy little girl who is now thriving with the right care and treatment. Even on days we know she doesn't feel 100 percent, she doesn't let it slow her down from playing, laughing and being a busy toddler. She loves her grandparents, Nanny Jane, her dog Samson, cheese and Elmo. Her outgoing personality and big brown eyes steal the hearts of everyone around her as she holds court everywhere she goes from restaurants to shopping malls. Her nonstop chatter and ability to recall the details of people she meets constantly surprises us. There is no doubt in our mind that she will achieve every dream we have for her and every dream she dreams for herself.

With research, possibilities are endless. Research gives us hope: the hope that our daughter will have the opportunity to dictate how she wants to live her life, not allowing cystinosis to dictate life for her. Knowing that she can achieve anything she desires, we hope she grows with confidence to pursue life with strength, grit and perseverance. We believe that no matter what life throws at her, she can rise up above it and keep moving forward.

We dream for a better tomorrow, and with ever-evolving treatments, our tomorrow grows brighter. With recent advancements in treatment and research, we believe Lily can achieve all this. But so much more can be done, needs to be done, and will be done. We plan to join the fight this year, to spread awareness and raise money for research. "Help Lily Blossom" will officially be standing strong with so many of you who dream courageously every second of every day.

LOLA'S STORY

On the Path to a Brighter Future

By Jim Long, Lola's dad

CHASKA, MINNESOTA

My wife Melissa and I have four children. Lola is 4 years old and is our youngest. She has two older sisters Ella, 13, and Ava, 11, and one brother, Jake, who is 8 years old. Lola's story started in much the same way as most other children... She was actually our largest child (in a group of "runts") by quite a bit. Lola was registering in the 60th percentile and 80th percentile in height and weight respectively until approximately 6 months of age, when she began to lose weight.

From 9 to 12 months, Lola continued to lose weight. At approximately 9 months the vomiting and the unquenchable thirst started. She had days where we were convinced there was more coming out of her stomach than was actually going in. On the day of her first birthday Lola went into Minneapolis Children's hospital with dehydration and failure to thrive. She would end up spending almost a month there.

The time Lola spent in the hospital was difficult, especially for her. Between the orders of no food and no liquid, the blood draws and fitful nights, we were watching her waste away before our eyes. After almost two weeks in the med surgery rooms, they moved Lola to the ICU. We were new to and struggling with all of the procedures she was being put through – many of which now seem almost commonplace. Lola was continuing to go downhill. It really hit us when the hospitalist told us we might want to begin preparing for a scenario where Lola might not go home.

When Lola was in the hospital three weeks (two in the ICU), they diagnosed her with Fanconi Syndrome. Fanconi Syndrome is a secondary condition resulting from kidney failure which is commonly found in children with

nephropathic cystinosis. The diagnosis ended up being the breakthrough we needed. Things moved fairly quickly from there. Soon after, Lola was diagnosed with nephropathic cystinosis. We began reading anything we could get our hands on regarding the disease. It was an emotionally polarizing experience; I am sure many parents of cystinotic kids have the same feeling – relief having a diagnosis but the relief is overshadowed by the uncertainty of the diagnosis.

We were set to begin our first dose of medication when we received a call from Lola's nephrologist. There was a clinical study beginning at Lurie's Children's Hospital in Chicago for a new drug, Procysbi®, that they wanted Lola to participate in. Remarkably, we received the call on the same day that Lola's Cystagon® (the legacy drug) arrived at our house after a delay caused by a mix-up with the pharmacy and insurance company. Had she received Cystagon®, she would not

have been eligible for the Procysbi® study because they wanted children who had not had Cystagon®.

In July 2013, we made our first trip to Chicago to be part of the study. Lola was going to receive her first dose of Procysbi® and we were anxious to start taking steps to slow the cysteine damage. However, because Procysbi® is a collection of very small beads, we were unable to push the medicine through the Lola's GJ feeding tube. We headed back to Minneapolis.

In August, we returned to Lurie's hospital. Lola's tube had been switched out for a larger G tube and the Procysbi® passed through just fine. We were excited that she was in the program and had access to doctors and researchers who were familiar with cystinosis. Her various medications still needed fine-tuning and Lola's weight was still low. But, we were excited to have what felt like a real starting point for progress. She couldn't communicate what she was feeling, but you could tell when her levels were off as she'd become very lethargic and not at all herself. The vomiting continued at a fairly heavy pace and we ended up having a couple of transfusions when her hemoglobin levels dropped very low.

For most of the fall of 2013 and into 2014 we would make bi-weekly trips to Lurie's. Although we thought we were seeing improvements, Lola's progress was slow. Around Christmas time we started to see a marked improvement. The frequency of vomiting dropped considerably and her labs seemed to stabilize – most of the time at least. We also noticed that she generally seemed to feel better. Her energy level was up and she almost always wanted to "play with the big kids." Lola continued in the clinical study for a year, and now is getting the drug Procysbi® commercially.





Cystinosis is a devastating disease in the ultra-rare category with only 2,000 cases diagnosed worldwide. All cells contain an amino acid called cysteine. Normally functioning cells have transport mechanisms that remove unneeded cysteine from inside the cell to be processed and expelled from the body. Children with cystinosis lack the ability to transport the unneeded cysteine out of individual cells, which allows the cysteine to build up and eventually form into crystals which end up damaging and eventually killing the cell. By most outward appearances, Lola and other children with her disease appear somewhat normal; however, internally every cell within their bodies is being ravaged by the disease. Even with medications, over time, cystinosis will destroy organs and core functions in their bodies.

There is currently no cure; cystinosis is considered a terminal disease. Lola and all children with cystinosis take a cocktail of medications throughout the day – most designed to try and stabilize chemical levels within their bodies. Lola takes 10 large pills each day and 12 liquid medications which are injected directly into her stomach through a G-tube. Additionally, cystinotic children develop crystals within their eyes which cause photophobia and can eventually lead to blindness.

Other than the frequent doctor appointments and scheduling around meds, we really don't treat Lola any differently from our other three kids. She is in preschool and loving it! She is also involved in gymnastics and was excited to begin dance this year. We feel very fortunate to have connected with the Cystinosis Research Foundation where 100 percent of the money donated goes directly to research. Being an ultra-rare disease, there is little to no funding outside of that raised by events like Curl for a Cure. With your help, we have hope that there will one day be a CURE!

It has always been very important to us that Lola has the same opportunities and experiences that our other children do. We have the same hopes and dreams for her as we have for her siblings. We wish for her to live her life to the fullest: to love, be loved and to one day have her own family. With all the new research and continued efforts from the Cystinosis Research Foundation and the cystinosis community, we feel that those hopes and dreams are becoming reality.





FINDING PEACE BETWEEN THE STORMS

By Valérie Talbot, Eva's mother
QUÉBEC CITY, QC CANADA

It's crazy how, in only a heartbeat, life can change so dramatically.

There was before. Life was easy, enjoyable. Now that we have grown accustomed to our new normal, we can't look back. We don't want to look back: seeing the pictures of sticky floors and stained clothes. Seeing her so thirsty without knowing why. Seeing her smile through her nausea in those before pictures, the pictures of that time when we didn't know. It hurts, even if it's actually easier now that we do know. The storm has passed. There will certainly be others, but since Eva's 16-month anniversary, the sun is shining again.

Eva was born at 8 pounds 3 ounces, in apparent good health. You know what that's like. We had a dream life with two beautiful princesses that completed our family! Eva was drinking ounce after ounce of milk; she was such a chubby baby. At around 4 months old, she started vomiting daily. She was drinking so fast that it all just came right back out. When she discovered water, she refused to drink any milk at all. Just seeing a bottle made her throw up, as did any attempts to feed her solid food. At 5 months, worried by her constant vomiting and seeing that she was beginning to lose weight, we consulted her doctor. At 6 months, she had her first blood tests. She had stopped growing and we had to find out what was wrong.

On that day, January 25, 2016, our whole life was turned upside down. We received a phone call: Eva's potassium level was too low. She was at risk for cardiac arrhythmia and we had to take her to the hospital right away. They would be waiting for us. A nephrologist was standing by when we arrived. The next day, she told us about Fanconi syndrome. A few days later, we heard her mention cystinosis for the first time. We were told so little and I was spending my entire evenings on the Internet, looking for more information. The doctor was telling me to slow down, that I was getting ahead of myself! But I needed answers; I needed to prepare myself. I needed to protect myself. I am so grateful for Eva's nephrologist. She already had a few young cystinosis patients and she knew right away what was wrong. No time was wasted!

We were released from the hospital 11 days later with a supply of phosphorus, potassium and Calcitriol, a tiny 6-month-old with a lifetime hospital subscription and many, many questions.

We were feeling so lost and I turned to social media for support. I found Cystinosis Family Canada, Parents of Children with Cystinosis, Cystinose France and more. I posted countless

questions to those groups and I can never thank enough all the girls who supported me. My English has improved since I began reading up on cystinosis! I have new English-speaking and French-speaking friends I can really count on, day or night. Every day, I see examples of how the cystinosis community is truly united. I am also happy that through this community, Eva has "cystas" her age. Despite the distance, I know that Charlotte, Odin, Brooke, Ellie and Eva will become fast friends.

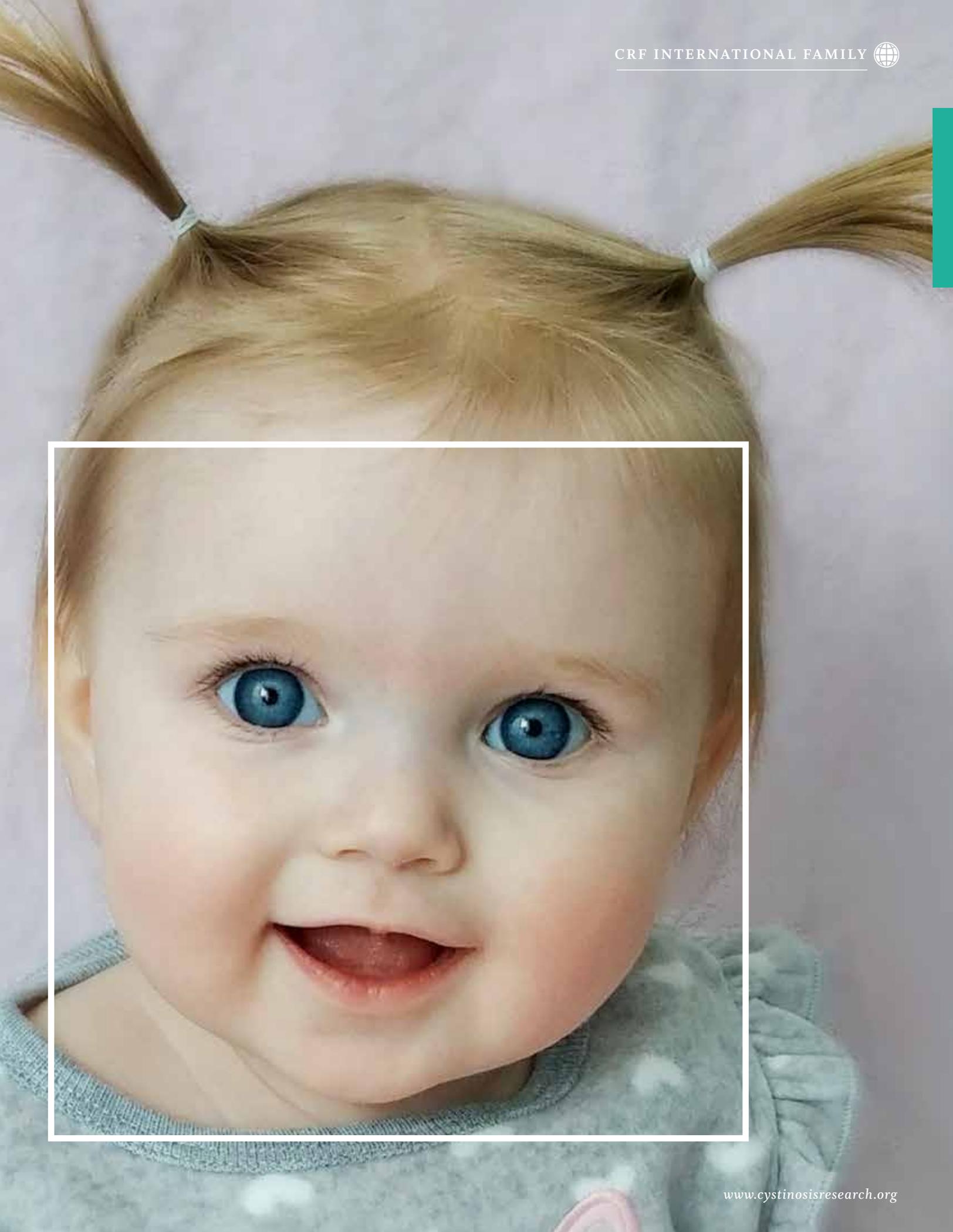
I would especially like to extend my thanks to the Stack family for their dedication and to the many researchers who spend their days looking for ways to make our lives easier. Thanks to everyone who answered my questions when Eva was so sick.

Eva has come a long way in the

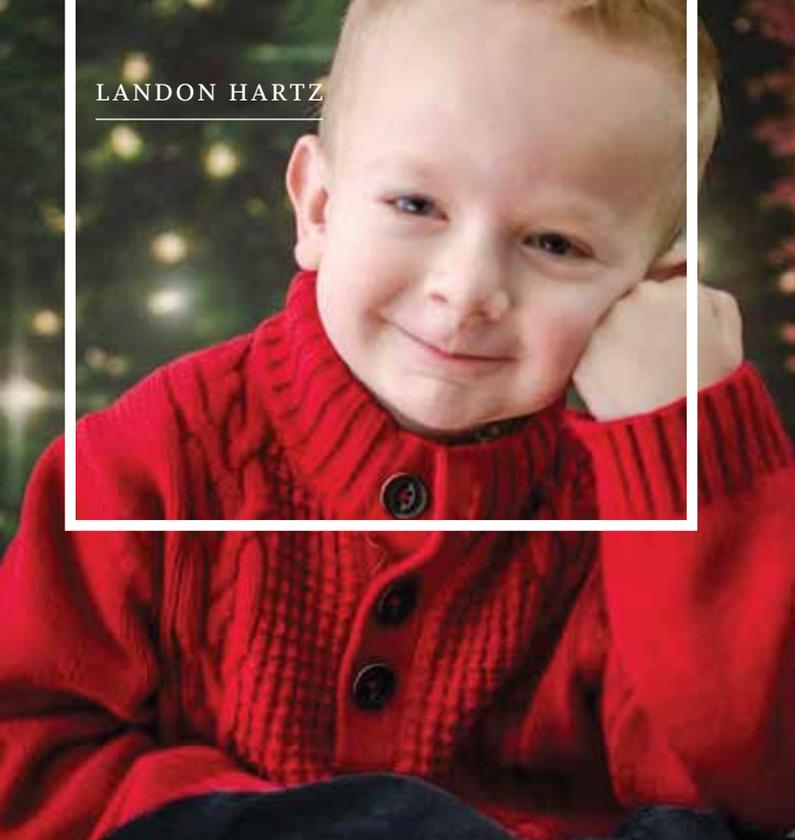
past year, despite six hospitalizations. She went from a weak little baby who refused any kind of food to an energetic toddler who beat her food aversion, who only rarely throws up and who is curious to taste (almost)

everything. Our lives are peaceful once more. We are happy once more. Eva sings, dances and always makes sure the house is as messy as can be!

Our lives are peaceful once more. We are happy once more.



LANDON HARTZ



Dear Me

5 YEARS AGO

Your sweet boy was just diagnosed with cystinosis. It won't always be so hard for you to spell. These 28 days you will spend with him in the hospital will be hard but you, Jimmy and Landon are very strong. You asked the nephrologist if he will be able to play sports and go to school. He will play sports and he will go to school. He was right in saying that life will be normal with a few bumps along the way. I know that you are totally overwhelmed by all of his new needs. You will make mistakes but this will become normal for all of you. It won't be sad. It just will be.

Dear Me

4 YEARS AGO

I told you. He has made a lot of progress but I know that it's still not easy. He still throws up a lot. It won't always be this way and will become a rare occurrence. He is eating a bit but is very selective and pockets his food before spitting it out. It will take baby steps but, in the not so near future, your grocery bill will sky rocket!

I know you are sad. You think that you won't have any more children. You will. You will have another little boy and he will not have cystinosis. When you receive the phone call that he doesn't have it, you will cry tears of joy and sadness. You will grieve Landon all over again.

Landon is in preschool now and has a really hard time separating from you. Trust that you both will know when he is ready for you to leave. One day he cried so hard, when you left, he threw up. You sat in the hallway, with him in your arms, waiting for the director to tell you if you had to take him home. You cried because you thought about all of the upcoming years when you will have to advocate for him at school. This also will get easier. One day he will turn to you and say "Bye Mommy!" and will walk into the classroom. From that point on, it will always be harder for you than it will be for him.

Love Letters

By Lauren Hartz, Landon's mom
PITTSBURGH, PENNSYLVANIA



Dear Me

3 YEARS AGO

Now you are the mom of two little boys and life is fun, chaotic and feels pretty normal. Landon is thriving. Less time is spent at doctor and therapy appointments and more time is spent with friends, family and at fun places. He loves school and is becoming more outgoing around people he doesn't know. You worry about having to answer questions Landon has about cystinosis and why he is different than his brother. He will ask and your answers will be loving and thoughtful. You can do this.

You went back to work and it has been great for you. Being a mom, particularly of a child who has special needs, has changed who you are for the better and it reflects in your work as a mental health therapist.



See Landon's community supporters on page 64



Dear Me

2 YEARS AGO

Landon is still doing so well. Your concerns are just like those of your friends who have kids. He is happy, healthy and his nephrologist describes him as a "normal kid who takes medication." Something is different in you now though. You notice that you often feel overwhelmed and anxious. Your body feels tense and your heart races. You wake up to give Landon his medicine at 2 a.m. and can't fall back to sleep for two or more hours. You hold a lot inside. You have taken pride in how strong you have been throughout this journey but powering through and staying in your head isn't working so well anymore. You will judge yourself for not really dealing with your emotions. You should give yourself a break. You were doing your best and you got through it. You needed to handle it how you did. It worked before and now it doesn't so you will become very curious about it and you will do some great work. It's your time, my dear. Your boys are doing great and your husband continues to be your rock. You will learn how to take better care of yourself and your life will change.



Dear Me

1 YEAR AGO

Landon is getting ready to go to kindergarten. You are telling yourself that he is ready but you are so worried. You talk to anyone who may have helpful feedback for you. You think about it and continue to think, think and think. And then one night you say to yourself "This is really hard for me" and you begin crying and cannot stop. Do you know what you are doing? You are staying in your head again because it's too hard to feel through this transition. Once you let yourself feel, it all comes together and you move through it. You talk to your Jimmy and together you decide to give him one more year before sending him to kindergarten. You will not regret this decision. You are afraid that you are making this decision because of you and not him. You're not.

Dear Me

TODAY

Not long ago you sent your baby to kindergarten. You are so grateful for how far he has come and how far you have come. This is a marvelous journey. He will do his best and so will you, and that will be good enough. It always has been.

With Love,
Lauren



“Let your dreams be *bigger*
than your fears,
your actions *louder*
than your words, and
your faith *stronger*
than your feelings.”

By Erin Little, Olivia's mom

PORT ELGIN, ONTARIO, CANADA

Our biggest fear of having a child with cystinosis is losing our child with cystinosis. Our dream is bigger than our fear and our fear is our driving force to fight for the dream. Our dream is simple and is like many others: To have a cure for cystinosis. We dream that Olivia becomes healthy and stays her happy, joyful self.

Olivia is 7 now and we are approaching our six-year “cystiversary.” Someone recently commented on the joy that Olivia exudes and wondered if she is truly happy; the answer is yes.

Olivia doesn't fully understand that she has cystinosis. If you told her she was sick, she'd look at you as if you were crazy and she would tell you that you are wrong. Our dream is that the cure comes before she ever has to make the wish for a cure, a wish made by Natalie that we are all grateful for.

Olivia is doing exceptionally well; her numbers are fantastic. She is tolerating her meds, she grew 11 cm in the past

Our dream is simple and is like many others: To have a cure for cystinosis.

year, her eyes are free of crystals and most importantly she is happy.

Growth hormones are new to Liv this past year, a decision we made that was not taken lightly. The thought of giving her daily injections broke our hearts. I would love to say it's easy for her, but I'd be lying. She takes it like a champ but watching her surrender her little body for a poke as she clenches her eyes shut and waits for the beep to signal it's over breaks our heart once a day, 365 days a year. Every day we think about how life is not fair for Olivia and we

wish we had cystinosis instead of her.

The thought of Olivia living a life of taking medications around the clock, every 6 hours, 365 days, 7 days a week, 52 weeks a year drives us into action. We fundraise to make our dream a tangible quest to find a cure. We spend our days thinking about fundraising and how we can spread awareness while trying to use caution while speaking about it in front of Olivia.

We choose to remain strong, fight for a cure, take action, dream big and have faith. Hope and faith drive us every day. Hope that we have a cure and faith that the universe has our back. At the end of every day we envision the cure, the celebration we will have when that day comes and the hopeful reality of 2,000 people worldwide living a life free of cystinosis.

Our dream is big but with faith, love and support we know it will soon be a dream come true.

own leaves



KADEN'S STORY: Making Each Day a Little Better than the Day Before

By Narlyn and Kahlied Thomas, Kaden's parents

PEMBROKE PINES, FLORIDA

We were packed and ready to go on our first family vacation, a seven-day cruise around the Caribbean. There were even going be T-shirts for us all: "Lil Papi" for Kaden, "Big Papi" for Kam, one for "Mom" and one for "Dad." There would be suits for the captain's dinner, swimming trunks and a first aid kit. Check, check and check. We even took the boys for a test drive around the living room in their new two-seater stroller. We had crossed our T's and dotted our I's but the Friday before the most awesome family trip ever planned, cystinosis happened.

For quite some time, Narlyn felt that there was something wrong with Kaden. She knew this because as she puts it, "We are in tune. Kameron and Kaden are the only two people in the entire world who know what my heart sounds like from the inside." She'd finally had enough of the lingering feeling that something was wrong with Kaden and could no longer take the wait-and-see approach. She could no longer hope he would want to drink more than just water, try a new formula in hopes that the milk would not be regurgitated, or wait for Kaden to reach the milestones that most babies reach. In just five weeks, Kaden had lost almost two pounds; none of the home interventions were working. Narlyn took him to the pediatrician for the third time in as many weeks, and finally the doctor agreed that Narlyn was right, and something more had to be done. The decision was made to take Kaden to the emergency room.

We found ourselves in a triage room at Joe DiMaggio Children's Hospital. A small room with a TV and a thin curtain separated us from all the other sick kids



and the chaos of the emergency room. Tests were run and after a few hours our nurse told us the team of doctors would be in to talk to us about Kaden. The team congregated outside our room and we heard someone say, "This kid's in bad shape." Of all the conversations we heard through that thin curtain, those were the only words that resonated. Denial and hope wouldn't allow us to believe that the "kid in bad shape" was our Kaden... But it was.

We were moved to a suite, and I foolishly reasoned that because our other son Kameron was admitted for asthma two years ago for two days, our stay with Kaden would only be for a day or two. The original problem seemed to be dehydration and a loss of protein and nutrients in his urine. It had been almost a month since Kaden had taken a bottle of milk, and when the doctors gave him a new formula, he gulped it down -- an entire 7-ounce bottle of formula was gone within a matter of minutes!

Excitement filled the room as we cheered and applauded our little man. We thought they had figured it out and he just hadn't liked the other formulas we'd tried at home. We hoped that the problem was solved and that we'd get our discharge papers,

go home and finish packing and set sail for the Caribbean. But we were wrong. The problems that caused the doctor to say that Kaden was in "bad shape" still persisted, even after he started drinking formula again. The protein and nutrients were still leaking, and it started to become evident that whatever was going on in his little body was going to take more than just a few days to figure out.

We were moved to the pediatric intensive care unit so that he could be better monitored. If Kaden could talk, I think he would say that the "bad people" were surrounding him and doing things like pricking his fingers and toes for blood up to four times a day. It was in those moments that we learned how loud Kaden could cry and how much it hurt us to see him that way. After two weeks of uncertainty, worry, NG tubes and ND tubes, needles, needles and more needles, we were finally able to put a name to the symptoms Kaden faced: that name was cystinosis. No one ever wants to hear their child has a genetic disease but it was comforting to have the weight of uncertainty lifted from our shoulders. We knew what it was and now, we knew what we had to do.

Kaden's medication regimen intensified. He could no longer take the volume of meds and meals through the tubes in his nostrils so a gastric tube was placed through surgery which meant more worry. We worried as we signed the waiver, as the anesthesiologist assured us that everything would be fine, as they rolled him away and as we nervously ate breakfast to escape the anxiety of waiting for the surgery to end. We worried until we finally got the call that it was a successful procedure and we

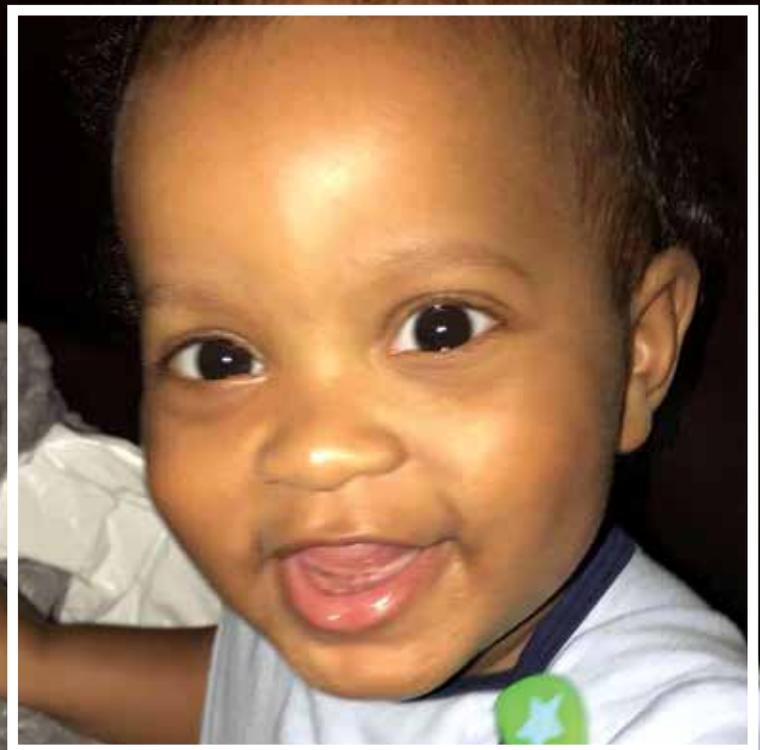
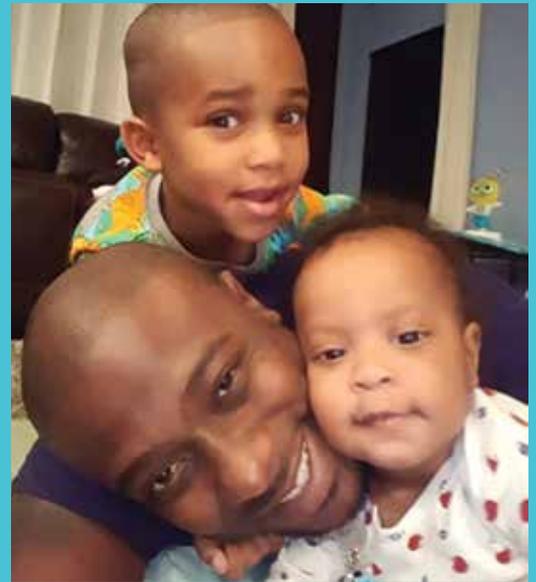


could finally see our little man, who was sleeping peacefully with a new hole in his tummy.

Gradually his numbers started to improve until finally the news came: We were going home! Forty days, seven room changes, 5,000 needle sticks (or so it seemed), tons of meds and countless procedures later, Kaden was back to where he needed to be. It was almost surreal moving out of our little temporary hospital apartment, packing up our belongings and saying goodbye to our tiny TVs and uncomfortable pull-out beds. The nursing staff, all of whom had become extended aunts, threw a nice going-away party in the hallway. Making our way to the exit we passed all of our old rooms, each holding within them memories we'll never forget – the pains, the joys, the laughs and the cries. But most importantly, within those seven rooms for those forty days, Kaden Amari exemplified everything that his name means: He was a fighter and he was strong.

We've been home for four months now and every chance big brother Kam gets, he reminds us that "We're a family now." Weeks of sleeping at Grandma's while we were in the hospital gave him a deeper appreciation of what that means. Kaden is doing a great job of taking back all the things that cystinosis wouldn't allow him to have a few months ago. He's crawling, walking, talking and all that other good stuff. It's been a challenge to say the least – the juggling of meds, the changing of sheets and clothes, the seemingly endless doctors' appointments and therapy sessions. But we've learned to take it day by day and, with the help of an outstanding parent support group, are making each day just a little better than the day before.

*Every chance
big brother
Kam gets, he
reminds us
that "We're a
family now."*





See Morgan's
"Sunglasses Day"
on page 60



Together

We're in This

Over the past 12 months, our family's cystinosis adventures have brought us to seven different events, spanning four states!

CALIFORNIA

CRF Annual Day of Hope Conference and Natalie's Wish Gala

DELAWARE

Chandler's Chance Christmas-Palooza

OHIO

Second Annual Mulligans Fore Morgan Golf Tournament and World Rare Disease Day Awareness (Sunglasses Day at Redwood Elementary School)

PENNSYLVANIA

Cystinosis Town Hall Meeting, Lots of Love for Landon Golf Outing, and Lots of Love for Landon Halloween Party

Over the years, we've formed irreplaceable bonds within our small (*but mighty!*) cystinosis community. We've witnessed the instant friendships that Morgan and our entire family make at fundraising events, meetings and conferences, and it inspires us to become more involved and to further expand our fundraising efforts.

Morgan has taken very big steps in the past year. She has become educated about her disease, has made cystinosis friends that she keeps in touch with through text and Facetime, and has continued to work hard at school. She's opened

By Jen Peachman, Morgan's mom
AVON LAKE, OHIO

up about her disease, and as a result, her best friends and teachers helped Morgan and her sister Maddie work the lemonade stands during our annual golf tournament. The Student Council at Redwood Elementary School even organized a "Sunglasses Day" on World Rare Disease Day this year to raise money for the Cystinosis Research Foundation in Morgan's honor.

Morgan has a great support system from her family, friends and teachers that makes her feel like we're all in this together.

It's hard to believe our daughter is turning 10 soon! Double digits. As she approaches another big milestone, we look back on the struggles she's had and the accomplishments she's made, and most importantly, we look to the future with hope.

Morgan: As Mom and Dad have always told you, we're in this together, sweet stuff!

We're full of excitement for what this year will bring for Morgan, as we watch her grow up all too quickly! Preparing for the Third Annual Mulligans Fore Morgan Golf Tournament is fun and exciting for the entire family. We also can't wait to join our cystinosis family for upcoming fundraising events.

The Cystinosis Research Foundation makes us hopeful, and the fundraising we do strengthens our hope. We know that every dollar raised brings us closer to a cure. Attending the events hosted by our cystinosis family – events that we've come to love (*and not miss!*) year after year – gives us hope.

After all, we're all in this together, and supporting one another fills our hearts and fuels our quest to find a cure.



**SAVE
THE
DATE**

SUNDAY, SEPTEMBER 17, 2017
THIRD ANNUAL
MULLIGANS FORE MORGAN
GOLF TOURNAMENT
BOB-O-LINK GOLF COUSE
AVON, OHIO





Ethan's ENDEAVOUR

By Amanda and Christopher Fenn, Ethan's parents

RUTHERFORD, NSW AUSTRALIA

Our son, Ethan, was born in September 2013 and everything seemed fine. As time went by and he was getting older, Chris and I realized that he wasn't thriving like a baby his age should: We took him to three different pediatricians, he had blood and urine tests run, and he was admitted to the hospital twice for dehydration, even though we were told that every child develops at his or her own pace.

At eleven months, Ethan was not crawling, talking or even sitting without support, and we knew something wasn't right. After pleading with the pediatrician to find out what was wrong, Ethan was admitted to the hospital again and went through three days of IV drips, seven blood tests, four urine tests, two stool tests and saw several different doctors. Finally, we were informed that Ethan's kidneys were leaking essential nutrients, and we were referred to the Renal Clinic at Westmead Children's Hospital.

On the 19th of September 2014, two weeks before Ethan's first birthday, I was told that he had cystinosis. As I had never heard of this disease before, I looked it up on the internet. As I read about it, I felt like my heart was being ripped out and my world was falling apart. It was one of the most difficult days for me. I held Ethan tight and told him we would get through this together.

Ethan is now on eight different medications every six hours which he must take for the rest of his life. It was so exhausting traveling back and forth to the hospital a few times a week, so when the doctor said that Ethan would need a gastrostomy tube to receive overnight feeds, I felt like I had been knocked down again. I had been doing all I could to help Ethan put on weight, but felt like I was not doing enough.

After 16 ½ months, Ethan finally took his first steps. Both Chris and I were there to witness this milestone, which really touched our hearts as Chris is normally at work and doesn't typically observe these events. We had all been waiting for the day when our little man had enough muscle tone in his legs to carry his body without assistance. We cried tears of joy and his siblings cheered him on when he did.

Ethan has thrived since getting the gastrostomy tube. We weigh in every night, and he loves to see every 100 grams

he gains and tell the family so we can all do a celebratory happy dance together. However, there are also those times when the scales tell a different story and his weight has decreased dramatically, which is upsetting for him.

Some people have grandiose dreams and shoot for the stars; Ethan is happy just to learn he has gained 100 grams after dinner. He is unassuming and happy all the time. He takes all his struggles in stride and shows no bitterness or jealousy toward others who don't have the same challenges. His younger brother, Morgan, who is not quite 2 years old yet, has already passed Ethan in weight and height. When Ethan learned that Morgan was not only heavier but also taller, he had nothing but pride in and happiness for his "little" brother.

We are sure one day he will ask questions like "Why me?" But for now, all he asks is, "Why not?" He wants to try everything and has a passion for life. When Ethan turned 3, he started taking karate with his three older siblings. It was an emotional day when he first put on his karate outfit and looked like a little ninja. Ethan has been so determined to keep up with his brothers and sister and is now just a few weeks away from graduating to his orange belt. He deserves all the happiness in this world and we know that cystinosis won't keep him down.

*Ethan is happy
just to learn he has
gained 100 grams
after dinner.*





2016 was a great year for me, but it did start out a little rough. On January 20, my parents and I headed to Duke University Hospital to begin the process of finding a new kidney. Due to sepsis, E.coli and age, my kidney had started to decline over the last few years. Now I'm not the type of person who gets very nervous or scared of much, but the thought of starting the whole dialysis process all over again after 24 years of a healthy kidney had me a little worried. I had heard way too many bad stories about dialysis and really started to dread it. My heart and prayers go out to those who deal with it on a daily basis. So, I did the whole Duke thing: numerous scans and tests and honestly was expecting the next few years to be a little difficult and maybe a lower quality of life than I was used to. But my faith and family had clearly gotten me through a lot, so I was as ready as I could be.

Due to that weak kidney, last spring and summer were filled with a lot of napping, blood tests and scans. But that wasn't going to get me down. Like previous years I really wanted to attend the Cystinosis Research Foundation Day of Hope family conference and for the first time I was able to bring my parents along! It was amazing as always and my parents were blown away! I really love experiencing it every year and seeing all the great families I've met along the way.



Bryan and his sister, Amelia, who donated her kidney to him on November 7, 2016

Bryan and Alex are set to be married October 21, 2017

My Best Year Yet

By Bryan Stout

CHERRYVILLE, NORTH CAROLINA

The weekend is always filled with many unforgettable “moments.” Last year was absolutely no different! Being there was about to change my entire life.

We all know cystinosis has its downfalls. But without cystinosis, I would never have met my perfect match in life. So, let me tell you a little about her: Alex is 29 and of course we have a lot in common. Her health is good, and she’s as beautiful on the inside as she is on the outside. We chatted a month or so on Facebook (she says I stalked her, but I promise I didn’t! lol) then finally met at the Day of HOPE. We really hit it off and I convinced her to come visit my family and me in North Carolina. We had a great time and that’s when we noticed we wanted a lot of the same things out of life. She must have liked something about the South because she came back about two months later!

We really had a great summer but the long-distance thing is difficult, and at that time I had all the tests going on for my kidney evaluation. My kidney function had finally dropped below 20 percent, therefore I was on the list and ready to get a new kidney! Alex was always there for me and I had started to realize she was truly something special.

I was now on the transplant list, which honestly was good news at that time, but I still dreaded the possibility of dialysis. Then I got the news that a fistula would be placed in early August, and it would be a two-part surgery (just my luck, lol) to be finished up in September. Once again, I was worried a little but I was ready. The surgery came and went without any complications and all the while Alex and I were trying to plan more trips to see each other!

A neat fact about Alex and me is that we are both hard-core Packer fans and she is from Wisconsin, so she planned a tailgating trip! The trip ended up being a blast. I met her family, and really enjoyed her showing me around Wisconsin! When I returned home, I never had to have the second fistula surgery. Now a lot of people say when you have a good story or something good happens it’s luck or coincidence. I don’t believe that. God answers prayers every single day! Even while waiting for a kidney my 2016 was continuing to be a great year for me. Alex moved to North Carolina in October!

As I said before, my first kidney lasted 24 years; it was now declining and I didn’t want to wait too long before I got a new one, but we all know it usually takes a while. Family members and friends helped spread the word and amazingly both my sisters ended up being a match! I simply can’t explain how that felt. Because my sister Maygen wasn’t a year post-partum she couldn’t donate. I had been on the list just over a month and my other sister Amelia called me. She said, “I’m a match.” I honestly said, “Huh?” I don’t know if I didn’t hear her or was probably just in shock. It was just unbelievable that my sister was going to donate a kidney to me very soon!

The surgery was scheduled for a month later. This was the reason I never had to have the second fistula surgery. So, from the time on the actual list until I received my new kidney was only about two months! Just three days after my kidney transplant, I truly felt like a brand-new person! The

crazy part is Amelia is terrified of needles but she took it all like a true warrior. She never complained, never wanted a pat on the back or anything. She is my hero and there’s truly not enough I can say about her. My other sister Maygen, who is a photographer, did a “transplant photo shoot” I guess you’d call it. We got a lot of local press out of it and even did a news story! Through all this, Alex never left my side and took care of me the whole way. Needless to say, by this time she was my girlfriend and also a great nurse!

What amazes me about the whole process was how my life had changed in such a short period of time. I probably say it too often, but I am blessed. I have had so many people whose lives were also touched

by transplants reach out to me after my story was in the news – it’s unbelievable. I still stay in contact with all of them! I just want to help others through support and do whatever I can for those out there who are still waiting for that organ they are in desperate need of.

It’s been almost four months since my transplant and I feel great! Duke actually told me not to come back for a checkup for three months; I also asked Alex to marry me in December of last year. On October 21, 2017 we are getting married. I couldn’t be happier.

ACTS 20:35: By all these things, I have shown you that by working in this way we must help the weak, and remember the words of the Lord Jesus that he himself said, ‘It is more blessed to give than to receive.’

“I am blessed!”

The Fight Continues



By Brian Sturgis, Henry's dad

SANDPOINT, IDAHO

As I write this article, I'm on a flight to Orange County for the Spring 2017 Cystinosis Research Foundation (CRF) board meeting. March Madness is bearing down on our family – and I'm not talking about basketball. March is the busiest time of the year for our family as it's the month we hold our annual 24 Hours for Hank fundraiser and CRF holds its annual family conference and gala for cystinosis research. This year they happen to be one week apart.

Both fundraisers bring about so much love and support from friends, family and community that enrich us with energy, determination, dedication and drive. We love the energy and hope everyone brings it to our cystinosis gatherings – there's really nothing else like it. We carry that feeling with us all year round and draw on the energy and support in hard times. 2017 marks the ninth year for 24 Hours for Hank fundraising events and although we are a lot more efficient at planning and executing the events, the added obligations take their toll. So of course, our dream is for a cure for cystinosis.

Henry is doing pretty well right now. He is in the fourth grade, working very hard to meet his goals. His main complaint is that his eyes hurt and bother him at school. The eye drops are hard to administer and tolerate every hour. His eyes always burn and itch; we look forward to better eye treatments that we know are on the horizon.

Henry has the natural "drive" to stick with things he wants to accomplish. He has been working really hard at school. He has a rigorous schedule and many tasks to do in his daily routine. We utilize the special education resource teacher and physical therapist at the school. He does physical therapy exercises every day and makes his way to the resource teacher every day for special learning tasks. His class is also an "experiential learning" class that develops a learning program based on each individual child's learning needs. Henry continues to use the orthotics he was fit for last year; they work well. The folks at Adapt Orthotics & Prosthetics in Coeur d'Alene, Idaho, make them to form to Henry's feet to help with his ankle and leg issues that are caused by bone and muscle development as a result of cystinosis. Henry wears them all day every day, in his regular shoes and ski boots. He is very used to them and relies on their support, especially when running around playing with his friends.

We had a really great winter up here in North Idaho, with lots and lots of snow. Henry loves to snow ski and participated in a couple programs at Schweitzer Mountain this year. Henry was a participant in the Junior Starlight NASTAR races, held every Friday night in January. His team won "Most Improved" over the four-week series!

Henry also participated in the Funatics ski program, which meets every Saturday from 9:30 a.m. to 3:30 p.m. to ski with a group of kids his age to work on skills and learning the mountain better. Henry also participated in our local parks and recreation's flag football team league last fall. He would play for 45 minutes with a 5 minute break and even got to play quarterback a couple of times.

Henry's dream is to have a cure for cystinosis. But he also dreams of other things for his life, as every child should. He dreams of being a veterinarian or a computer programmer also known as a "coder."

This time of year when we feel like we can't put any more on our plate, we think about the Stack family. They have been at this fight much longer than we have, and have never wavered. Nancy and her team work tirelessly year-round to help our children. We pray that they will always have the energy and drive to continue. Their energy is contagious and motivates us to continue the fight!

I know tomorrow night as I fly back to Sandpoint after the board meeting, I will feel energized by all of the information that I received and the conversations I've had with families who are faced with the same challenges.

Thank you, Stacks, for the inspiration and energy! The fight continues!

*He dreams
of being a
veterinarian
or a computer
programmer
also known as
a “coder.”*



Our Escape to the City



Dear Friends,

Last year was a rough one. My cancer treatment and related risk-reducing surgery, and Jenna's surgery on both knees in December had us confined to the house for months. Kevin suggested we get out of town, and through a (very generous) friend, we had access to an apartment in San Francisco. Kevin, Patrick, Jenna and I escaped for three nights of fun in a city only 90 minutes from home, yet worlds away!

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



Jenna and Patrick on Alcatraz



SAN FRANCISCO, 2017

Even at our most carefree, cystinosis is present in all we do. The four of us are so used to living with or around cystinosis that we have to consciously consider the extra work we take on as a family in order to “take a vacation.”

We departed Thursday eve with suitcases packed for the weekend, a storage-size bin of waterproof and spare bedding to protect our host’s mattresses from the kids’ incontinence, twelve doses of medications: 340 pills total, for our three-day adventure, plus a set of crutches and a wheelchair for Jenna, who chooses which to use based on how far she has to travel. In the back seat with the kids are four quarts of water, which will just get us to our destination 90 minutes away, with a couple potty stops in-between. This family isn’t afraid of a nature pee. Faster. Cleaner. We can make it off and back onto the freeway in 3 minutes! We arrived, re-made the kids’ beds, gave them their meds, and hit the sack. Eleven hours’ sleep is best for Jenna, who is growing and healing, and who we have to work hard to keep healthy. A missed dose of meds, low blood sugar or too little sleep can put her in a tailspin.

FRIDAY: ALCATRAZ

We awoke to sunny skies for our 9:30 departure to Alcatraz. Laundry. No washer and dryer at the apartment. The kids stripped their beds and I looked up Fluff & Fold businesses on Yelp. Kevin made friends with the owner of Five Star Cleaners on Russian Hill. She took our basket of soaked bedding, and said it would be ready after 5 p.m. We would eat breakfast on the way to the Ferry Terminal. Medications must come after a meal, so I put the kids’ morning doses in my purse.

Alcatraz was so intriguing, and we all enjoyed the audio tour. We boarded the ferry to the island via the handicapped line, as Jenna was in her wheelchair, and we toured the island via tram, as the hills and the distance to the tour site were more than either of the kids would endure. We had two water bottles that we refilled at every water

fountain we came across. At noon, Kevin realized that we had forgotten the meds in my purse, so on the ferry ride back we purchased water and gave the kids their morning meds, five hours late. Jenna returned to the apartment with her dad for some down time. Patrick joined up with friends from his sixth grade class for Pokémon hunting and a tour of a WWII cargo ship docked at Fisherman’s Wharf, enjoying a purely fun afternoon with friends. As the boys pretended to shoot the guns at the bow of the warship, the other mothers and I watched and considered that boys only 5 years older than they were once at the helm of this ship. In the back of my mind I considered a “perk” of cystinosis: Because of the disease, Patrick wouldn’t ever be drafted. We picked up our laundry from Five Star Cleaners (\$45!) and enjoyed dinner at Fisherman’s Wharf before heading back, re-making beds, taking meds, and tucking in.

SATURDAY: FARMERS MARKET, CHINATOWN, NIKE STORE & THE TONGA ROOM

Five Star Cleaners was ready for our fly-by on Saturday morning. We dropped off the laundry and drove to the Farmers Market at the Ferry Terminal. Patrick and I agree that given one meal for the rest of our days, it would be the Chilaquiles at this market. We were a little more punctual with morning meds, giving them to the kids in the middle of the Ferry Building at 9 a.m. We once again divided: Kevin took Patrick to Nike for new shoes, and Jenna and I headed for Chinatown to shop for candy and trinkets at the bazaars. I pushed Jenna in her wheelchair the one mile from the Embarcadero to Chinatown, half of it uphill. If you can conjure a visual of this, and add to it my tendency toward speed walking and a fairly vigorous shimmy in one of the wheels of the chair, you would see why Jenna and I were in hysterics for most of this quick journey. We did find trinkets, ginger candies, pretty painted fans and a flower clip for Jenna’s hair. We enjoyed lunch and hailed



a taxi, making sure the car was one that could accommodate a wheelchair, and went back to the apartment for a nap.

The Finale: Saturday dinner at the island-style Tonga Room. We ordered and ate slowly, the kids enjoyed special drinks, dramatically delivered in carved-out pineapples. Live, island-style music was played from a stage, which was floating in a pool in the middle of the room, tiki torches all around. The kids talked about their futures, their hopes and dreams, where they wanted to live, and the friends they would never lose touch with. To watch them was to know that they were two very happy kids. As we departed the restaurant, Jenna claimed it was the best night of her life.

The future feels more and more uncertain as the kids grow and we manage days filled with the responsibility and heartache that come with cystinosis. In a parallel universe, our trip to the city might have included a lightly packed car, and dry bedding each night. Jenna and Patrick might be seen skipping up the hill on Alcatraz to take their tour, experiencing no more thirst than their dad and me. There would be no pills to take to stay alive, no upset stomachs from pills, and no regrets from giving pills too late. I dream one day, we will find a cure for cystinosis, and that our children’s road will merge with the one in that parallel universe. That road might find Jenna and Patrick, at 20, exploring Europe, with only a toothbrush and a change of clothes in their backpacks: having no thoughts of thirst, pills, or pain.

With our deepest gratitude for your commitment to our dreams,

Teresa Partington (Kevin, Patrick & Jenna)

MUSIC
FOR
MARY

An Incredible Success

By Melissa Head, Mary's mom
RAINIER, WASHINGTON

The Fourth Annual Music for Mary took place on February 10 at the Tacoma Sportsman's Club in Puyallup, Washington. It was both an exciting and emotional night!

More than 170 guests attended the event that featured our favorite local crooner, Chris Anderson. This was Chris' third year singing for Music for Mary and he is always a hit!

The night included a silent auction with more than 50 silent items, a live auction, a delicious Italian dinner catered by Pellegrino's and 25 gorgeous desserts for the dessert raffle.

Melissa's uncle Gary was back to MC the event with his wife, Debbie, better known as our own "Vanna White," to showcase the live auction items. Gary kept the crowd entertained with a game of Heads and Tails as well!

Toward the end of the evening, Shannon Keizer took the stage to speak about her life with cystinosis. Shannon's resilience, strength and compassion for people and life were motivation for everyone to push on despite what circumstances might hinder them. Shannon had the whole crowd moved to tears.

To end the night, a slideshow to "Fight Song," (inspired by last year's Day of Hope where Rachel Platten performed), was presented to the crowd. So many generous hearts gave money during the Raise the Paddle: almost \$3,000!

We were honored to have five other cystinosis families in attendance! The Keizer family from Michigan, Shank family from Oregon, Patterson and Seachord families from Washington and the Suetta family from California.

The night was incredibly successful, raising awareness and over \$21,000 for the Cystinosis Research Foundation!



See Mary's
lemonade fundraiser
on page 63



Reaching a Comfortable Stride

By Marcu Alexander, Hadley's mom
BOISE, IDAHO



My hope and dream is that in another five years Hadley will no longer have cystinosis.

It's been five years since Hadley was diagnosed with cystinosis. Our lives have changed so much since those early days. The first year was definitely the most scary and challenging for us. The unknown was terrifying and the future seemed bleak. There were new medications which required several dosage changes, constant blood draws, daily vomiting and G-tube surgery. She wouldn't eat and literally survived on massive amounts of whole milk fortified with Duocal to get the necessary fats and carbohydrates her body needed. There were several hospital stays, IVs and sleep was scarce. Our new world felt isolated and overwhelming. How could we possibly maintain this new lifestyle while providing our daughters with the love, care and support needed to develop tiny humans into decent members of society? How would we ever be able to trust anyone else to provide the care Hadley needed to survive? Would big sister, Stella, grow up feeling left out and less important because she didn't have a rare disease? We were plagued with guilt and scared at the thought of what the future held.

As the months and years carried on, life began to feel more manageable. Blood draws weren't as frequent; there were fewer medication changes; Hadley began to eat and the vomiting slowed down. We trained caregivers, family members and friends how to administer meds through her G-tube

which allowed us a chance to go out on dates and regain some balance in our relationship. There was more to life than cystinosis! We did have setbacks along the way with hospital stays and the addition of new medications. However, those instances became less daunting as our confidence grew and we gained more control.

As we enter our fifth year of managing this disease it feels like we've hit a milestone! Life is much easier today and we have hit a comfortable stride. Hadley's health has been consistently good and there are times we forget she even has an illness. She recently threw up and my first thought was that she must have a tummy bug. It's been so long since she's vomited that cystinosis wasn't even my first thought.

Hadley started kindergarten in the fall and has had an incredible year so far! She quickly acclimated to school life and has made many new friends. Her growth has been outstanding, which is attributed to her voracious appetite. We once worried she'd never eat food and now she will eat three tacos in a single sitting. She lives for spicy kimchee and loves all seafood!

Hadley has recently taken on a larger role in her own care. She administers her own afternoon medications every day and fills her own water bottle when she's thirsty. Blood draws are no longer an issue and she

happily sits in the chair alone during the procedure knowing her reward will be some stickers and a prize once it's over.

We don't take this point in our journey for granted. We know she will once again face hard times, but we feel more prepared for when that time arrives. Nighttime is still a huge challenge for us with several diaper and bedding changes along with multiple refills of milk in her sippy cups. She is sensitive to the fact she must wear a diaper to bed at age 6, even on the nights she wakes up to go potty. This will continue to be difficult as time goes on and she begins to have sleepovers with friends. But we will face those issues just like we have in the past. We're fortunate to have support and guidance from fellow cystinosis parents and the Cystinosis Research Foundation.

My hope and dream is that in another five years Hadley will no longer have cystinosis and we'll be able to look back with appreciation on the road we traveled to reach the cure!

How Cystinosis *Inspires Me*

My name is Tina Flerchinger. I am in the seventh grade and I am 13 years old. I go to Cornerstone Christian School. I was born with cystinosis, but at the time my parents didn't know I had it. When I got older I didn't eat, I would throw up all the time, and I drank so much water. My parents had two other children before me, (Nichole, 9 years older than me; and Catherine, 8 years older than me), so they knew that this was not normal. My parents tried their hardest to find out what was wrong with me, but doctors didn't know.

One day, when I was 17 months old, my doctor found out I had cystinosis. At 18 months, I started taking my meds, but since I wouldn't eat I got my first G-tube. I took all my meds and food through that G-tube. I always had a backpack on my back so I could carry all my tubes and pump in it. As I got older, every so often I would get a new G-tube. I remember the pain it was to take a tube connected to the G-tube out and then press a new one back in.

By Tina Flerchinger
CLARKSTON, IDAHO

When I was around 5 years old, (I was still taking the Cystagon®, which makes you throw up all the time and not feel good), my mom was taking the capsules off my Cystagon® so that it could go in the G-tube. Well, when she was doing that, she looked away and I put the pills in my mouth because I loved to be sneaky when I was little. My mom turned around and realized that the pills were gone. She asked if I had taken them and I just laughed and giggled. Eventually she found out I had taken them. We found out I could take pills, so we slowly started giving me pills and eventually when I was 6 years old I got my G-tube taken out and had stitches.

As the years went on I was doing better, but every so often I would go to the ER at the hospital because I would have a cold or a fever, and it would be hard because I take lots of meds and would throw them up and would have to retake them, or if I had a sore throat it would hurt to take all my pills. Now I take 62 pills a day, two syringes a day, eye drops five times a day, and a shot every night.

My parents, (Mark and Denice), and my sisters have been so helpful and caring to me all these years. Without them and God I could not make it. I have become a stronger person because of cystinosis. Cystinosis inspires me to live my life, not take it for granted, but to live it fully for God! It has inspired me to follow my dreams and become a doctor. I want to be a kidney doctor for kids when I get older because I love helping others and I would tell them it would be OK and tell them how far I have come and that they will make it far in life too!

I love playing with my friends at school and my cousins. I play the piano and I love playing tennis with my friends. My favorite subject is science and I love doing Legos. One of my all-time favorite things to do is snorkeling and to go boogie boarding in the ocean. I am very eager for 2017! It is a huge year for cystinosis. We may have found the cure, and the first adult patients are doing treatments with it this fall. Even though I have been through obstacles in life, I am not afraid for the future, because God is with me and is always by my side. ❤️



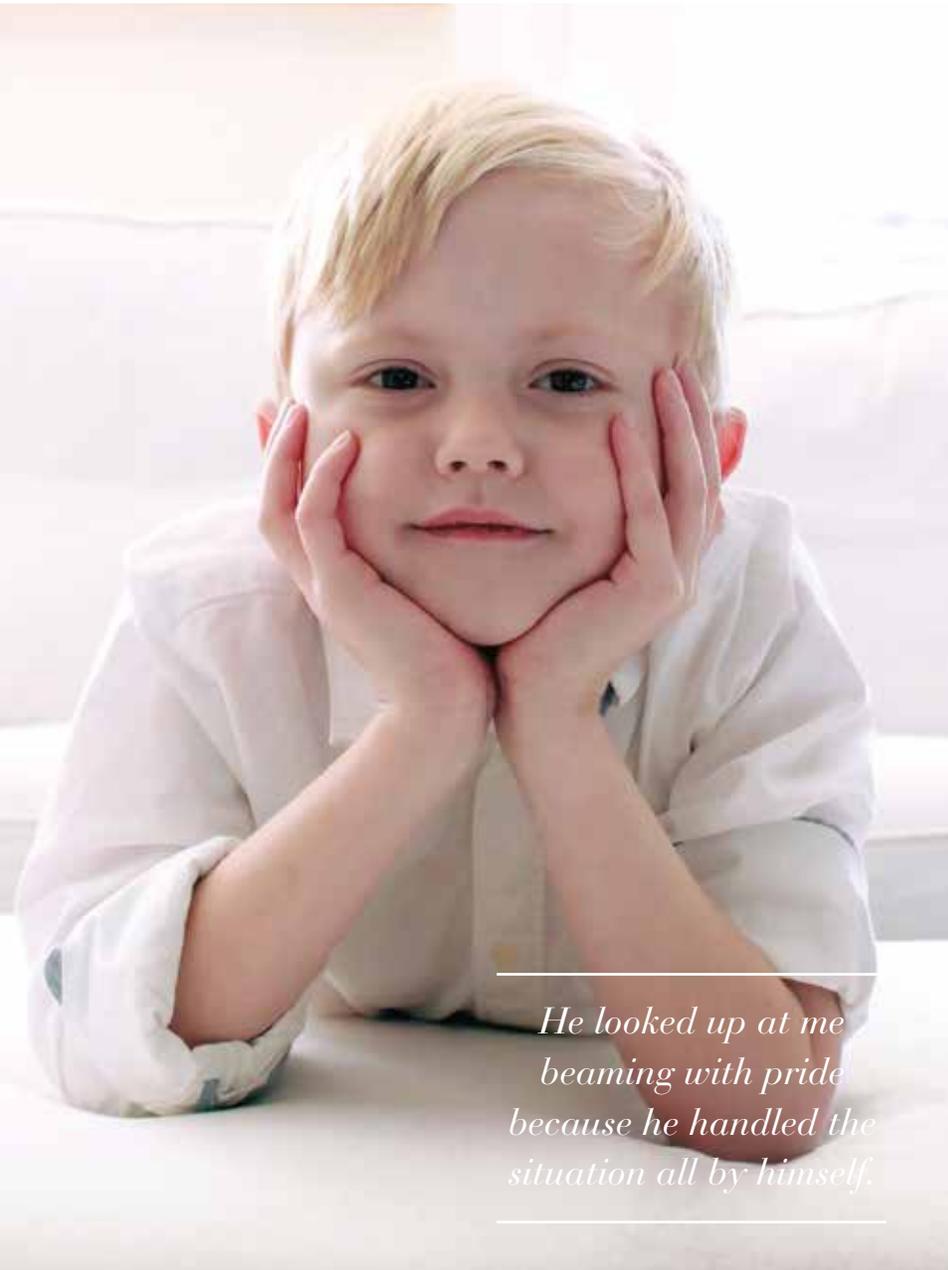
*I am very eager
for 2017! It is
a huge year for
cystinosis.*



*See Tina's
community
supporters on
page 66*

'Mmmmm . . .

By Ashton Jenkins, Sam and Lars' mom
SALT LAKE CITY, UTAH



*He looked up at me
beaming with pride
because he handled the
situation all by himself.*

There are times in my life when I feel myself living on autopilot.

My sons and I live constantly by a checklist, which includes (in no particular order): Give Sam and Lars Procysbi®, administer eye drops, give Sam electrolytes, give more eye drops, throw in morning laundry, schedule nephrology appointments, clean carpet where Lars threw up, give eye drops, go to ophthalmology appointments, get lab results, don't forget to refill potassium at the pharmacy . . . and the list goes on. In the midst of the chaos I sometimes forget that our entire family is affected by cystinosis in many more ways than just physically.

Some of my hardest moments as a mother are when my children come home from school and tell me about an incident where a kid has made fun of them because of the way they smell. Cysteamine produces interesting smells that I've had other kids describe to me as "fishy," "sour milk," "dirty," and my favorite, "Mmmmm, like spaghetti dinner." Though most kids mean no harm, it is still difficult to know that my kids have to experience this scrutiny regularly. Since I can't be there to protect or defend my kids all day, every day, we work on phrases to say. For instance, "It's the smell of my medicine that keeps me healthy" or if the kid is teasing, a simpler "Well, that was rude" sort of response. I often ask Sam how these experiences make him feel. He says, "Sad." But he also says he is "happy" he has cystinosis because of friends like Henry and Tina that he gets to play with at CRF conferences.

Spaghetti Dinner'

During one of his first soccer practices, Sam playfully pulled the bottom end of his soccer shirt over his head revealing his G-tube. A boy on his team noticed and immediately asked what that “thing” was. Sam answered confidently that it was a G-tube that helps him take all of his medicines because he has a genetic disease called cystinosis. He then looked up at me beaming with pride because he handled the situation all by himself. The words “genetic, disease, and cystinosis” aren’t words you’d want a young kid to know but I was very proud of him.

Stephen and I are very open about cystinosis with our kids and do our best to answer their questions. This past year Lars discovered that his friends do not have a medication schedule. With wide eyes, he told me they don’t swallow pills at all! He thought this was an amazing discovery and wanted to know when he could stop taking pills. It pulled at my heart strings to explain to him that medicine was essential to keeping his body healthy. Thankfully he has an easy-going personality and is a champ at taking his pills.

Although our schedule can feel both monotonous and chaotic, I’m learning how to pause and process the frustrating emotions that cystinosis can impose. We are impressed with our two boys and how they tackle their daily demands of cystinosis.



Lucky Number

Could our lucky streak continue? My wife and I are both healthy, as were our four children and the first five grandchildren when they were born. Our sixth grandchild gave us a scare, and there was a great deal of concern that he might need a pacemaker installed at birth. His mother received numerous treatments during pregnancy to prevent any damage to her son, and when he was born he went straight to intensive care for cardiac monitoring. After several days, it was determined that he could return to his family. He is now 3 years old, happy and healthy. He does have an irregular heartbeat, but it does not stop him from doing all that he wants to do.

When our seventh grandchild was born, everything looked great. Our little leprechaun, Preston, was born on St. Patrick's Day, 2014. He needed a little oxygen to get started, but that's not unusual. When he finally got to be with our daughter and son-in-law, Ciara and Shane, it looked as though we had dodged another arrow. He grew and developed just like any other infant, and all was well until just before his first birthday.

The problems seemed simple enough at first, and Preston's doctors said that his issues were normal in the early development process. But Ciara and Shane could sense that something was wrong and felt that the doctors were overlooking something. The constant constipation, frequent vomiting, excessive urination – were these really normal? We all felt that there had to be something causing all of these symptoms. Our daughter would call my wife and me daily to discuss Preston's condition, and the concern in her voice was palpable. She and my wife spent hours on the phone talking, crying, listening to and supporting each other. We all knew that something was wrong with Preston. Our challenge was to find a doctor who could tell us what it was.

The turning point came during our family reunion in May of 2015. Preston had just turned 1, and it had become apparent that he was not growing. His appetite and thirst were insatiable. His pediatrician prescribed mild laxatives for his constipation, and told our daughter not to worry as he would soon outgrow it. It was unusually hot the day of the reunion and Preston struggled to stay hydrated. Ciara left to take him home, and they landed in the emergency room. Once again, the doctors treated his dehydration and ran labs, but then released him without any further testing or information.

Although it may sound like I am bitter toward the medical community, I am not. As a former paramedic, I have a bit of medical training; what is taught in medicine is that if you hear hoof beats, look for horses. The pattern of doctors' visits, lab work, and no diagnosis continued for another month. Finally, our daughter had enough. She made an appointment with Preston's pediatrician and asked us to come and support her while she "got mean." Anyone who knows Ciara can attest that she is mild, soft-spoken, and doesn't like confrontation. However, when it came to Preston's health, she was a different person. She was as strong as I had ever seen her and approached the situation in a way that only a mother of a sick child could understand.



I went with Ciara to the doctor's office while my wife stayed home to tend to Preston's older sister, Mikelle. I'm always proud of my daughter, but that day was special as I saw a part of her that I had never seen before. She made it clear to the doctor that we were not leaving until we had some answers or until we had found someone who could give us the answers.

The doctor left the room and came back in with two big medical research books. He spent an hour in the room with us, asking questions and looking through the volumes. We finally came to the conclusion that Preston needed to see the doctors at Primary Children's Hospital. Still thinking that this was a gastrointestinal problem, Preston's doctor called the hospital while in the exam room with us, and made an appointment for him with a pediatric gastrointestinal specialist. The doctors at Primary Children's are fantastic; they have weekly meetings to discuss their difficult cases, and it was during one of these meetings that it was determined that Preston should see a nephrologist. From that point, it only took a couple of weeks until we had the diagnosis. The good Lord will bless all of these doctors one day.

I'm sure that you have all had those times in life when something happens that you won't ever forget, even if you wanted to, as the details are indelibly stamped in your memory. This is how I felt when I received the sobbing phone call from my daughter. All she could mutter between the tears was "look up cystinosis and call me back." I did exactly that, and my heart sank as I read through website after website. How could it be this bad and, more importantly, how would I muster the strength to call my daughter back? Getting our strength from each other, my wife and I called Ciara back together. While we had not yet gotten Preston's diagnosis, we all knew after our research that he had cystinosis. Our phone conversation

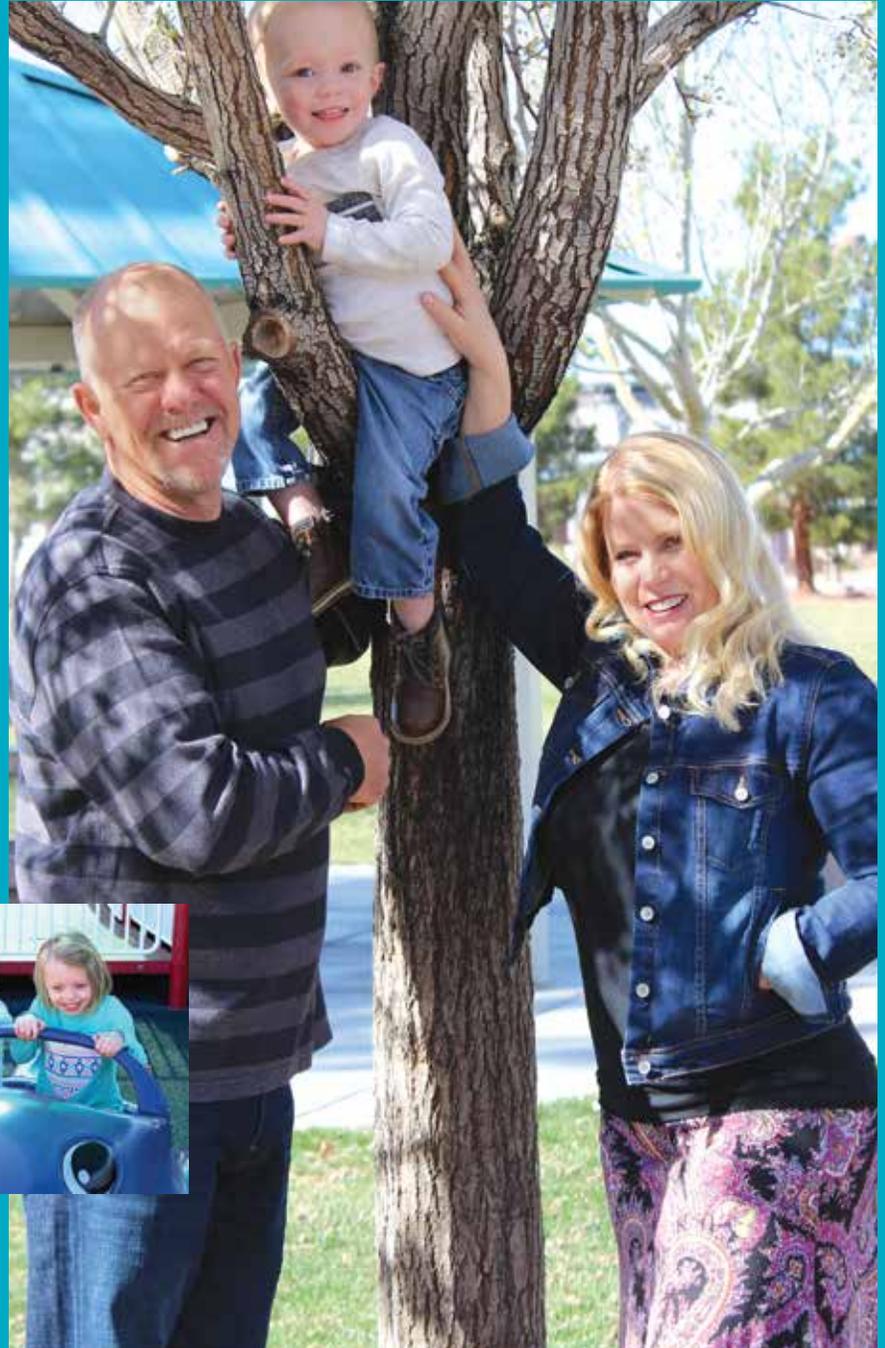
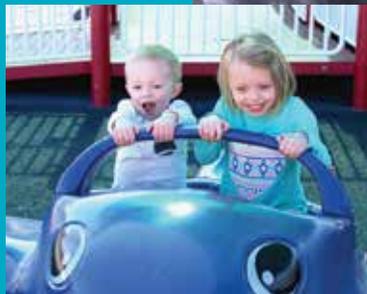


By Tim Hartman, Preston's grandpa

LEHI, UTAH

quickly turned into one of hope. There were new medications that could not only improve his quality of life, but could also give him time – time to find better treatments, time to find a cure. Time, love and hope were all we had.

While this article is entitled “Lucky Number Seven,” you could argue that luck has not been on Preston's side. Preston would argue that he is very lucky. He is lucky to have been born into a home with two loving parents, to have a fantastic big sister, and to have an extended family that loves him and cares about his happiness. Preston is lucky to have been born at a time when the Stack Family and the Cystinosis Research Foundation are working tirelessly to find a cure. Preston has no idea that he is sick. He is a normal, happy, adventurous 2-year-old. Everyone around him knows of his challenges, the potential outcomes and potential end of the story. But, because of Preston and his marvelous spirit, we also know about hope. We know how precious time is and how easily it can be wasted. We now know that the story of Preston's life is a work in progress with many potential outcomes. His life will take many twists and turns, but he has his mom and dad, his sister, Mimi and Papo, Grandma and Grandpa, aunts, uncles, cousins, his faith, and a wee bit of “The Luck of the Irish.” He's our little leprechaun, our lucky number seven, Preston.



Because of Preston and his marvelous spirit, we also know about hope.



IN LOVING MEMORY AND CELEBRATION OF

Weston Tschannen

BROOKFIELD, MISSOURI

Weston was born on January 21, 1993, along with his twin brother, William. He was diagnosed with cystinosis at the age of 17 months and took on this disease with a fighting spirit. He never let cystinosis define who he was. It was just a daily annoyance that he had to endure in order to enjoy all that life had to offer him. A lover of sports, cooking and music, Weston demonstrated his strength of character by finishing fifth in the 103-pound division of the Missouri State High School Wrestling Championships in 2009. Winning the last match of his 10-year commitment to the sport, Weston defeated his opponent with only 10 percent kidney function. Weston suffered complete renal failure at age 16. Receiving the loving gift of a kidney from his mother in 2009, Weston experienced a new lease on life until that kidney failed in 2014, when he began dialysis for the remainder of his life.



Weston further exemplified his determination to overcome life's challenges by completing his last year of college while undergoing daily dialysis treatments. A year ago, Weston started experiencing grand mal seizures and would later be diagnosed with epilepsy. This was particularly challenging because he could no longer drive. After further testing, Weston was put on two different seizure meds this past March and responded well to the medication. He graduated in May from the University of Missouri in Columbia with a Bachelor of Science degree with an emphasis in restaurant and hotel management. Most recently, following his graduation from college, Weston began interning with the research department of Raptor Pharmaceuticals, assisting in the development

of programs and town hall meetings to provide help for sufferers of chronic illness.

Weston had a wonderful spirit and such a zest for life! He accomplished so much in his short 23 years and we are so proud of him and so blessed to call him our son. Unfortunately, Weston's life was cut short on August 28, 2016, due to a very high potassium level and another grand mal seizure. His twin brother, William, who is an EMT, did everything he could to save his brother's life but God had other plans for Weston. Devoted to his family, Weston was a loving, funny, loyal companion to his many friends and members of his north Missouri community. Weston will be terribly missed by our family which also includes his brother Ryan and his wife, Anne; his sister, Ally, and her fiancé, Andrew; and by the many people he touched and those who fought by his side. He taught us so much throughout the years in dealing with cystinosis and continues to teach us how to live life to the fullest and enjoy every minute!

Weston's twin brother, William, and their closest friends organized a fundraising golf and blackjack tournament on the weekend of May 13 to help show support for the cystinosis community. Any and all people were invited to attend, with the golf tournament beginning in the morning on Saturday and ending the day playing blackjack. All proceeds will be donated to the Cystinosis Research Foundation. For information, please don't hesitate to reach out to William at wtsch121@gmail.com.

Loving family, The Tschannens

LEARN • SHARE • COLLABORATE

2018

CRF International Cystinosis Research Symposium

SAVE THE DATE

THURSDAY, MARCH 1 + FRIDAY MARCH 2
2018

WE ARE PROUD TO ANNOUNCE
OUR 2018 SYMPOSIUM CO-CHAIRS



Corinne Antignac, MD, PhD



Stéphanie Cherqui, PhD



Julie Ingelfinger, MD

LOCATION

ARNOLD AND MABEL BECKMAN
CENTER OF THE NATIONAL ACADEMIES
OF SCIENCE AND ENGINEERING
100 Academy, Irvine, California 92617
www.nationalacademies.org/beckman/

INFORMATION

Contact Foundation President NANCY STACK
at 949-223-7610 or nstack@cystinosisresearch.org



BY INVITATION ONLY
CYSTINOSISRESEARCH.ORG

CRF Joins OnPAR to Accelerate Research Accessibility

The Cystinosis Research Foundation is participating in a new paradigm for gaining access to new research through OnPAR, a public-private partnership between Leidos Life Sciences and the National Institutes of Health.

Through this opportunity, CRF will gain access to promising, but unfunded NIH research proposals that may complement the extensive research already being funded through the foundation.

OnPAR links high-quality research applications with potential funding matches among participating organization members,



according to their specialized areas of interest.

“OnPAR presents a great opportunity to fund promising and highly-scored research applications to sustain the pipeline of researchers, support new ideas and move research from bench to bedside for the cystinosis community,” said Nancy Stack, president and founding trustee of the Cystinosis Research Foundation.

Martin Dueñas, Director of Health Research Management practices for Leidos Life Sciences, the company that originally launched OnPAR, welcomed CRF to the program. “OnPAR seeks to provide a global pipeline of research projects to revolutionize the scientific funding environment and accelerate discovery to improve lives for people impacted by cystinosis,” he said. For more information, visit

[HTTPS://ONPAR.LEIDOSWEB.COM](https://onpar.leidosweb.com)

MARY LEONARD, MD, MSCE

CRF-Funded Researcher Appointed Chair of Pediatrics at Stanford



Clinical research expert Mary Leonard, MD, who with Dr. Paul Grimm led a CRF-funded clinical study

into musculoskeletal disease in children and adults with cystinosis, has been appointed chair of pediatrics at Stanford University and physician-in-chief at Lucile Packard Children’s Hospital Stanford and Stanford Children’s Health.

Dr. Leonard has received high praise from colleagues and trainees for her thoughtful leadership and inspiring vision for the future of pediatric research, education and patient care. Her research into the effects of chronic diseases on nutrition, physical function and bone health has extended into studying those with cystinosis, resulting in the first

comprehensive evaluation of bone structure and muscle strength in children and adults with cystinosis.

“This is an exceptionally exciting time for Stanford pediatrics,” she said. “The growth of our clinical and research programs and the new initiatives in precision health are providing us with unprecedented opportunities to shape the future of pediatrics.”

A 1989 graduate of Stanford University School of Medicine, Dr. Leonard returned to her alma mater in 2014 after spending 25 years at the Children’s Hospital of Philadelphia and the University of Pennsylvania, first as a resident and fellow and then as a faculty member. She is a member of the American Society of Clinical Investigation and the Society for Pediatric Research.

Your voice matters. Register at the CCIR website today:
WWW.CYSTINOSISREGISTRY.ORG



HELP THE RESEARCH COMMUNITY LEARN MORE ABOUT CYSTINOSIS AND ITS COMPLICATIONS.

The development of new treatments can be a lengthy process, and there are two steps that patients can help accelerate: the collection of valuable disease information and recruitment of volunteers to clinical trials. Your participation in a patient registry can achieve this acceleration.

The Cure Cystinosis International Registry (CCIR) is the most far-reaching cystinosis patient registry in the world, with 576 registrants from 44 different countries, including 63 new registrants in 2016. Thanks to those who have completed the online CCIR medical survey, the impact of cystinosis is

becoming more apparent, and researchers can better understand the concerns of patients and their families. Earlier this year, an expanded CCIR medical survey was introduced. The expanded survey is intended to capture richer, more detailed information about cystinosis that experts say is lacking in the medical literature and is necessary for advancing therapies.

If you have not yet had the opportunity to take the expanded survey, please do it now. It only takes a few minutes.

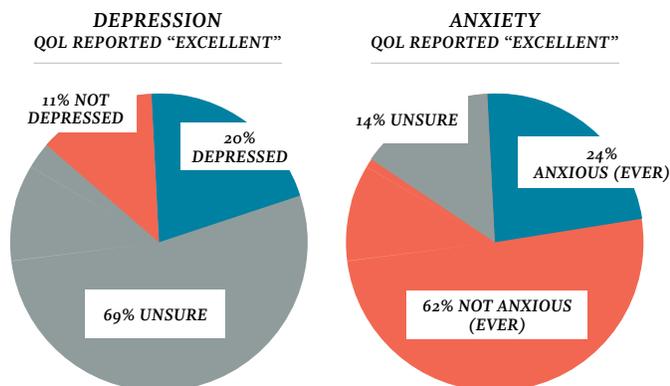
Your feedback on current care and treatments for cystinosis is critical for the identification of research areas to focus on in the future.

RECENT RESULTS

REPORTED QUALITY OF LIFE (QOL)

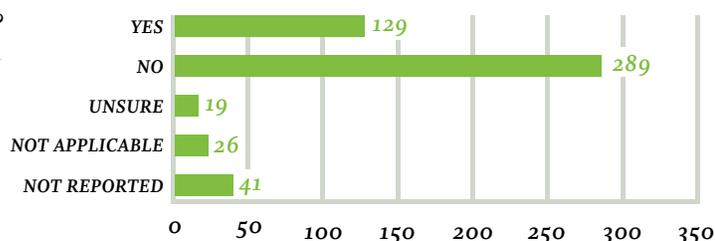


Even though a large portion of patients report good or excellent quality of life, a significant percentage of those reporting excellent quality of life suffer depression or anxiety.



HAS THE PATIENT EVER HAD LEARNING DIFFICULTIES?

In one of our neurological questions, we learn that most patients do not have learning difficulties. But about 1/3 of respondents have learning disabilities of some kind.



**SAVE
THE DATE**

NATALIE'S WISH TENTH ANNUAL
FORE A CURE GOLF TOURNAMENT

at the world-class Pelican Hill Golf Course in Newport Beach
Monday, October 16, 2017

CRF FORE A CURE RAISES \$441,000

BRINGING THE TOURNAMENT TOTAL TO MORE THAN \$3.0 MILLION

The Ninth Annual CRF Natalie's Wish Fore a Cure Golf Tournament held on October 17, 2016 at the Pelican Hill Golf Club was another tremendous success. Over 200 community and business leaders attended the record-breaking tournament raising over \$441,000 for cystinosis research! Our energetic Chairman Vince Ciavarella led a powerhouse golf committee who helped gather major sponsors and underwriters for the sold-out event. And our dedicated group of volunteers welcomed golfers and assisted throughout the day.

"The exceptional generosity and support of our friends and the community has enabled CRF to make significant advances in the treatment of cystinosis. CRF-funded researchers at institutions around the globe are making important

breakthroughs toward the cure. We are honored to be a part of the progress to help Natalie's wish become a reality by contributing to cystinosis research," said Vince.

After a rainy start to the day, the sun appeared with clear blue skies to complete a perfect round of golf. Golfers then gathered for a cocktail reception and barbeque dinner outside at the golf pavilion. The festivities included a silent auction followed by a dynamic and spirited live auction. The tournament's reputation as the "the best in Orange County" was confirmed as golf sponsorships were sold out several months in advance and because of our committed and energetic golf committee, most of the underwriting opportunities were sold by tournament day.

Since 2007, the Fore a Cure golf tournaments have contributed more than \$3,064,585 to cystinosis research and our quest for a cure. Our dedicated committee and volunteers, most of whom have been involved since the first tournament, are the driving force behind the Fore a Cure tournament and their efforts helped make this year's event the most successful ever.

Because of the dedication of our donors and sponsors the Fore a Cure Tournament earned Top 5 honors again from the Orange County Business Journal's Charity Event rankings, placing fourth with most revenue raised. We are grateful for your continued commitment to make Natalie's wish become a reality, "To have my disease go away forever."



Contact Zoe Solsby to become a sponsor
(949) 223-7610 or zsolsby@cystinosisresearch.org


THE RESORT AT
PELICAN HILL
NEWPORT BEACH

TOGETHER, WE ARE **One**

MORGAN PEACHMAN

STUDENTS SPORT SHADES TO SHED LIGHT ON RARE DISEASE AWARENESS

To raise awareness for Rare Disease Day held on February 28, 2016, the Student Council at Redwood Elementary School in Avon Lake, Ohio, hosted “Sunglasses Day” in honor of Morgan Peachman. Students and staff showed their support by wearing their sunglasses and donating \$1 or more to cystinosis research. As the word spread, another elementary school in the district joined the cause! Funds raised were donated to the Cystinosis Research Foundation in honor of Morgan. Not only was this a fun and creative way to create awareness, the event was a success, **raising \$597 to help find a cure!**



JOSHUA CLARKE

JOSHUA'S JOURNEY OF HOPE

On Sunday, February 5, 2017, Team Josh – consisting of Cynthia Fritz (aunt), Abigail Fritz (cousin), Rose Clarke (mom), and Jonathan Clarke (brother), met in Huntington Beach, California, to participate in the Surf City Half Marathon. This was no ordinary run, it was a mission to raise funds and awareness for cystinosis research and a CURE!

Over the previous months, Team Josh raised awareness and solicited donations via GoFundMe using Facebook, church bulletins, and word of mouth. The response was better than expected with contributors from across the nation, from California to New Hampshire.

Together, **they raised more than \$5,000** for cystinosis research at this event, meeting their fundraising goal. Joshua and his family are grateful to their friends, family and all the wonderful people who contributed to this important cause. Team Josh had fun running and Joshua had fun watching!



1 PURPOSE. 1 JOURNEY. 1 CURE.

GABBIE STRAUSS



GREAT FRIENDS OFFER ENDURING SUPPORT

Gabbie Strauss has wonderful friends who continue to support her year after year. One of them, 9-year-old Delaney Laking, hosted another lemonade fundraiser in honor of her friend Gabbie. The lemonade stand was a success and **raised more than \$106!** Gabbie is grateful to have a dear friend who supports her and cystinosis research.

In other news, Moserfest had yet another successful summer gathering on August 27, 2016. The rain didn't stop friends and family from enjoying the festivities, food, music and donating to cystinosis research. **The event raised more than \$2,400.** Gabbie Strauss and her family are thankful for the continued support of Jeff and Mel Moser who planned the event.

MARY HEAD



LEMONADE STAND PRODUCES SWEET RESULTS

Mary Head's longtime friend Mason Smith had been wanting to do a lemonade stand in honor of Mary for years. But living out in the country in Puyallup, Washington, is not an optimal spot for such an idea so this presented a problem.

When Mason's grandma was going to have a yard sale, they realized her house would generate traffic and be a perfect place for the stand. When asked what he was going to do with the money, Mason replied, "Give it to Music for Mary!"

Mason spent two days selling homemade lemonade and sweet tea, **raising \$67** for the Cystinosis Research Foundation.

Mason's generous heart no doubt comes from his parents, Matt and Megan Smith. Megan is on the Music for Mary committee and puts hours and hours of work into getting sponsorships and donations. The Smiths are always among the first families at the Music for Mary event to help set up and are always the last to leave after they help clean up. Thank you Mason and the Smith family!

TOGETHER, WE ARE **One**



KEEGAN MANZ

LETTER-WRITING CAMPAIGN FOR KEEGAN

In December, Brad and I wrote a letter asking for people to show their support by making a donation to the Cystinosis Research Foundation. One of our neighbors posted the letter on our neighborhood subdivision's website. We were humbled by the donations that came in from our new neighbors – many of whom we have not met yet. We are so grateful for our friends, family and community's dedication to finding a cure for cystinosis. Keegan's letter-writing campaign was a huge success **generating almost \$8,000!**

2016 was quite the year for Keegan! He moved into a new town, started preschool and mastered the scooter. He works hard to do what comes easy to others. Our priority this year has been to make sure Keegan has the right support to feel successful in different environments. Keegan has been thriving in preschool because of the support of many wonderful teachers and a therapist. He has made many friends and is trying new things each day at school!

6TH ANNUAL HALLOWEEN PARTY

The Hartz Family held their 6th Annual Lots of Love for Landon Halloween Party Fundraiser. Three hundred people were in attendance and the guests enjoyed a catered dinner, treats, music and dancing with Mr. Matt Colella who generously donated his time and services. The costumes were wonderful making it difficult for the judges to choose winners from 5 different categories! The 50/50 raffle, twenty door prizes and thirty Chinese Auction prizes gave guests a lot of opportunity to leave with something. **\$11,450 was raised and donated to the Cystinosis Research Foundation.** Thanks to all who attended and for all those individuals and businesses that supported this event!

LANDON HARTZ



SOUTH UNIVERSITY IN GEORGIA DISPLAYS SUPERPOWERS IN COMPASSION

Landon Hartz has a team of superheroes supporting him in his fight against cystinosis.

His cousin, Danielle Vernillo and her husband, Mike, coordinated several events through South University's 2017 Spring/Summer SouthCares Committee including a bake sale, a donation drive to wear jeans to work, and a "Make Me Super" competition in which a member of each team dressed up as a superhero.

Landon got to pick the winner of the superhero contest. But with a **total of \$1,302 raised for cystinosis** from these events, they were all winning heroes in his book.

1 PURPOSE. 1 JOURNEY. 1 CURE.

BAILEY DEDIO



HIS HOPES, HIS DREAMS AND A CURE

2ND ANNUAL BAILEY BELIEVES FEST

The Orange County community rallied around Bailey DeDio by supporting the 2nd Annual Bailey Believes Fest **raising \$7,700 for cystinosis research**. Special thanks to Josanna Charity of Doggie Stylin' Grooming and Lucky Johns Bar and Grill for their efforts to organize and host another successful fundraiser to fund cystinosis research.

CALEB GOWAN



CHILI FOR CALEB'S CAUSE

The Spirit of Hope Church in Mesa, Arizona, recently hosted a fundraiser, Chili for Caleb's Cause, organized by Jim and Karen Gowan in honor of their grandson,

Caleb Gowan. The Gowan family is grateful for the support of church members, the church youth group TAG, and their community including Trivent Financial who contributed to the event activities and fun. Karen commented, "We are so thankful for the successful event which **raised \$1,900 for cystinosis research**. And we are blessed by the faith of our family and all members at the Spirit of Hope Church. Thank you!"

TANNER EDWARDS



STORM
MEN'S SHOP

THE GENDRON FAMILY

The Storm Men's Shop of Fort Collins, Colorado recently hosted a Fashion For Charity runway show in honor of Tanner Edwards to benefit the CRF and cystinosis research. The event organized by Traci Gendron and her son, Tanner, provided guests with an opportunity to view the latest in men's fashions while supporting the cystinosis community. The event **raised \$1,540 for cystinosis research!** The store is committed to donating profits from sales to cystinosis research. Please visit them online and shop for the latest in men's fashions while helping the cystinosis community! www.StormMensShop.com

TINA FLERCHINGER



BREAKFAST FOR DINNER

Miss Lewiston USA 2016, Brooke James, organized Tina's Breakfast for Dinner on October 6, 2016. Brooke's aunt, Becky Tatsey, owner of Waffles n' More, generously said yes to hosting the event and donating proceeds to Tina's Hope for a Cure. Guests were served "Half the Farm" and Mimosas by the James family and the friendly Waffles n' More staff. It was a very fun evening, and the food (as always) was fabulous. **An amazing \$2,600 was raised for cystinosis research!** The James family have generously supported cystinosis research since Tina's diagnosis. We are grateful for their continued love and support.

ERIK KELLY

CHILDREN'S FUN-RUN BENEFITS CRF RESEARCH

Students at School of the Good Shepherd in Victoria, Australia, showed up in force last year to support their fellow classmate, Erik Kelly, who has cystinosis. Working together, they hosted an Adidas School Fun-Run to educate the community about the disease and **raised an impressive \$5,833.05** for cystinosis research. Donations were collected for every lap completed by participating students. That was motivation for everyone to do their best. "The school raised a generous amount for a little school," said his parents, Maria Pekli and Daniel Kelly. "We feel very lucky and grateful for being a part of this community."



TINA FLERCHINGER

STELLA & DOT PARTY

Sherry Seeh, a Tina's Hope for a Cure board member, held a Stella & Dot Jewelry & Accessories Trunk Show at her home in Clarkston, Washington on August 19, 2016 to raise money for Tina's Hope for a Cure. Mariel Schneider, a Star Stylist with Stella & Dot in Edmonds, Washington, approached Sherry about having a fundraiser party after reading about Tina's story and cystinosis. Of course, Sherry jumped at the opportunity to hold a fundraiser, having friends come to her home to purchase beautiful jewelry and accessories, all the while helping Tina's Hope for a Cure!



With Mariel living on the far western side of the state and Sherry living on the far eastern side of the state, the logistics didn't pose a problem. Sherry displayed samples of the products, took purchase orders, while Mariel did the "virtual" presentations on Facebook. While the ladies enjoyed shopping, they were particularly focused on hearing more about Tina's current health updates and the status of cystinosis research. The party was a huge success! The attendees were motivated to do anything that would help raise money for Tina's Hope for a Cure. Together, Sherry & Mariel donated 100% of their earnings to "fund a cure." **Raising \$610.00** wasn't bad for a small in-home party!

In the process of finalizing the event, Sherry joined Stella & Dot as an Independent Stylist. She plans to use her business as a way of donating funds to local nonprofits. Of course, she will be having another Stella & Dot trunk show to support Tina's Hope for a Cure! Every dollar DOES count!

SAVE THE DATE

Thursday, April 19, 2018 — thru — Saturday, April 21, 2018

The Island Hotel, Newport Beach, California



learn | share | laugh | celebrate

Day of Hope

CYSTINOSIS RESEARCH FOUNDATION
FAMILY CONFERENCE

Cystinosis families from around the world are united in their quest for a cure and have funded 151 multi-year research studies which have led to a new FDA-approved drug and new discoveries about cystinosis. We will celebrate our community at the Day of Hope conference and will renew our efforts to work together in our quest for the cure.

Learn, share, laugh and celebrate for three exciting days at the 2018 Cystinosis Research Foundation Day of Hope Family Conference and Natalie's Wish celebration at The Island Hotel in Newport Beach, California.

The Day of Hope conference will include sessions led by CRF-funded researchers and clinicians. Topics include:

- Updates on stem cell and gene therapy
- Novel eye treatments for corneal cystinosis
- Kidney disease and transition to adult care
- Updates on muscle wasting and myopathy
- Bone and muscle clinical trial update
- Day-to-day living with cystinosis

A partial list of confirmed speakers include:

- Ghanashyam Acharya, PhD
- Sergio Catz, PhD
- Stéphanie Cherqui, PhD
- Paul Grimm, MD
- Morgan Fedorchak, PhD
- Robert Mak, MD, PhD



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



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.

Saturday, March 4, 2017

CURLING TOURNAMENT FOR LOLA LONG

Chaska, Minnesota

Contact Melissa Long: melismahan@yahoo.com



Friday & Saturday, March 24-25, 2017

24 HOURS OF SCHWEITZER SKI EVENT, HENRY STURGIS

Schweitzer Mountain, Sand Point, Idaho

For information visit www.24hoursforhank.org



Saturday, May 13, 2017

GOLF TOURNAMENT FOR WESTON IN MEMORY OF WESTON TSCHANNEN

Kansas, Missouri



Friday, May 19, 2017

PARS FORE PRESTON GOLF TOURNAMENT IN HONOR OF PRESTON LUKE

The Legacy Golf Club, Henderson, Nevada

Contact Ciara Luke: ciarahluke@gmail.com



Saturday, May 20, 2017

HOPES & WISHES - JAKE KRAHE

Weymouth Country Club, Medina, Ohio

Contact Amy Krahe: ajkrahe@gmail.com



Friday, June 2, 2017

6TH ANNUAL LOTS OF LOVE FOR LANDON GOLF OUTING IN HONOR OF LANDON HARTZ

Black Hawk Golf Course, Beaver Falls, Pennsylvania

Contact Jimmy Hartz: lotsoflove4landonCRF@gmail.com



Thursday through Sunday, September 8-10, 2017

**SACRAMENTO CAPITAL CUP GOLF TOURNAMENT
JENNA & PATRICK'S FOUNDATION OF HOPE**

Sacramento, California

Contact Teresa Partington: tjpartington@sbcglobal.net



Saturday, September 9, 2017

**FUN RUN IN HONOR OF SAM AND LARS JENKINS,
PRESTON LUKE AND BRANDON WALDRON**

Draper, Utah

Contact Ashton Jenkins: ashton.bee@gmail.com



Friday, September 15, 2017

**HEARTS FOR HADLEY EVENT
IN HONOR OF HADLEY ALEXANDER**

Boise, Idaho

Contact Marcu Alexander: hearts4hadley@gmail.com



Saturday, September 16, 2017

**SWING, SHOOT & LIV GOLF CLASSIC
LIV-A-LITTLE FOUNDATION**

Saugeen Golf Club, Port Elgin, Ontario, Canada

Contact: Erin.little@livalittlefoundation.com



Friday, September 29, 2017

**SWING FOR A CURE GOLF TOURNAMENT
24 HOURS FOR HANK**

Stoneridge Golf Course, Blanchard, Idaho

Contact: information@24hoursforhank.org



Friday & Saturday, October 13-14, 2017

**7TH ANNUAL SWING AND BLING EVENT
JENNA & PATRICK'S FOUNDATION OF HOPE**

Swing Golf Event - Catta Verdera Country Club

Bling Event, Railyards in Sacramento, California

Contact Teresa Partington: tjpartington@sbcglobal.net



Monday, October 16, 2017

**CYSTINOSIS RESEARCH FOUNDATION - NATALIE'S WISH
TENTH ANNUAL FORE A CURE GOLF TOURNAMENT**

Pelican Hill Golf Club, Newport Beach, California

Contact Zoe Solsby: zsolsby@cystinosisresearch.org



The Impact of CRF Research

CELLULAR AND/OR MOLECULAR STUDIES OF THE PATHOGENESIS OF CYSTINOSIS

39 GRANTS

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



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 Levchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

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

26 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

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THE SCRIPPS RESEARCH INSTITUTE,
LA JOLLA, CALIFORNIA

Holger Willenbring, MD
UNIVERSITY OF CALIFORNIA,
SAN FRANCISCO

CURE CYSTINOSIS INTERNATIONAL REGISTRY (CCIR)

1 GRANT



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

GENETIC ANALYSES OF CYSTINOSIS

4 GRANTS



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

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

Elena Levchenko, MD, PhD
UNIVERSITY HOSPITAL LEUVEN, BELGIUM

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

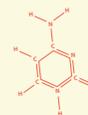
Minnie Sarwal, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN FRANCISCO

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 Levchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

THYROID

1 GRANT



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



AREAS OF RESEARCH FOCUS *and* GRANTS SINCE 2002

KIDNEY RESEARCH

18 GRANTS



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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 ZÜRICH, INSTITUTE OF
PHYSIOLOGY, ZÜRICH, SWITZERLAND

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

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

Tara McMorow, 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,
GOSSÉLIES, 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, BONE

6 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

13 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**

7 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

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

LEADERSHIP. GUIDANCE. COMMITMENT.

The Scientific Review Board (SRB) is composed of leading cystinosis scientists and experts from around the world. Members are actively involved in the grant-review process, evaluating and analyzing all research proposals that are submitted for potential funding, and advising the CRF on the scientific merit of each proposal.



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Laboratory of Hereditary Kidney Diseases
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PARIS, FRANCE

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Martin Konrad, MD

Department of General Pediatrics
University Children's Hospital
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*Thank you for your dedication to
the global cystinosis community.*

67 Published Studies

by CRF-funded researchers including the one below since the Fall 2016 issue of *Cystinosis Magazine!*

The Renal Fanconi Syndrome in Cystinosis: Pathogenic Insights and Therapeutic Perspectives

Published December 2016 in *Nature Reviews Nephrology*

by **Stéphanie Cherqui, PhD**

UNIVERSITY OF CALIFORNIA, SAN DIEGO

and **Pierre Courtoy, MD, PhD**

DE DUVE INSTITUTE, BRUSSELS, BELGIUM

CRF GRANTS ARE LEVERAGED BY MILLIONS **\$12,214,460**

NEW \$5.2 MILLION GRANT AWARDED TO STÉPHANIE CHERQUI, PhD



CRF-funded researcher Stéphanie Cherqui, PhD, at the University of California San Diego was recently awarded a two-year \$5.2 million grant from CIRM (California Institute of Regenerative Medicine). The grant will allow Dr. Cherqui and her team to do the pre-clinical work needed to demonstrate that the potential treatment is both safe and effective, paving the way for FDA approval of clinical trials in the near future.

Dr. Cherqui's goal is to take blood stem cells from people with cystinosis, genetically modify them to remove the mutation that causes the disease, then return them to the patient. The hope is that the modified blood stem cells will create a new, healthy, blood system free of the disease.

CRF issued its first grant to Dr. Cherqui in 2007 to explore the possibility of a stem cell treatment to cure cystinosis. CRF and Dr. Cherqui have worked closely together over the years to ensure the research moved forward. "This award is the result of pivotal work funded by the Cystinosis Research Foundation," Dr. Cherqui said. "I share this success with CRF, who has always been so supportive of me since the beginning of this project." Since 2007, Dr. Cherqui has been awarded over \$10.6 million in grants from other grant institutions. Today, we are closer than ever to a cure for cystinosis. We are extraordinarily proud of Dr. Cherqui and thankful to her for her dedication to the cystinosis community.

Collectively, CRF researchers have been awarded \$12,214,460 in additional grant funds by other institutions as a result of "seed" money provided by CRF. The discoveries made by CRF researchers benefit the greater scientific community and assist other more prevalent diseases and disorders. By funding the cystinosis research community, CRF donors are effectively helping millions of people with other diseases.

TOTAL 2016 GRANTS FUNDED: **\$2.79 million** FUNDING 17 RESEARCH PROJECTS IN FIVE COUNTRIES.

2016 FALL CRF Research Grants Funded

CHARACTERIZATION OF MTORC1 SIGNALING IN EARLY PATHOGENESIS OF CYSTINOSIS

Corrine Antignac, MD, PhD, Principal Investigator
Imagine Institute, Paris, France

\$310,000 – 2-year grant
(February 1, 2017 – January 31, 2019)

MOLECULAR TRAFFICKING REGULATORS OF DYNAMIC ORGANELLES IN CYSTINOSIS

Sergio Catz, PhD, Principal Investigator
The Scripps Research Institute, La Jolla, California

\$256,795 – 2-year grant
(February 1, 2017 – January 31, 2019)

EVALUATION OF CTNS -/- MICE PROTECTION BY ORAL SUPPLEMENTATION WITH BASIC AMINO-ACIDS: FOCUS ON KIDNEYS

Pierre Courtoy, MD, PhD, Principal Investigator
Christopher Pierreux, PhD, Co-Principal Investigator
De Duve Institute, Brussels, Belgium (PI)

\$209,344 – 2-year grant
(February 1, 2017 – January 31, 2019)

ALTERNATIVE THERAPIES FOR NEPHROPATHIC CYSTINOSIS

Francesco Emma, MD, Principal Investigator
Laura Rega, PhD, Co-Principal Investigator
Bambino Gesù Children’s Hospital, Rome, Italy

\$229,660 – 2-year grant
(February 26, 2017 – February 25, 2019)

BIOPHYSICAL STUDY OF CYSTINOSIN AND PQLC2

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

\$75,000 – 1-year grant
(February 26, 2017 – February 25, 2018)

LESSONS FROM THE FRUIT FLY. HOW CELLS COPE WITH CYSTINOSIN DEFICIENCY

Matias Simons, MD, Principal Investigator
Imagine Institute, Paris, France

\$91,553 – 1-year grant
(March 1, 2017 – February 28, 2018)

PHARMACOKINETIC EVALUATION AND OPTIMIZATION OF CYSTEAMINE PRECURSORS

Vincent Stanton, Jr., MD, Principal Investigator
Patrice Rioux, MD, PhD, Co-Principal Investigator
Thiogenesis Therapeutics, Inc., San Diego, California

\$153,900 – 1-year grant
(March 1, 2017 – February 28, 2018)

SLEEP DISTURBANCE AND MEMORY FUNCTION IN NEPHROPATHIC CYSTINOSIS

Doris Trauner, MD, Principal Investigator
University of California, San Diego, California

\$62,895 – 1-year grant
(February 1, 2017 – January 31, 2018)

2016 SPRING *CRF Research Grants Funded*

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

Sergio Catz, PhD, Mentor

Jinzhong Zhang, PhD, Research Fellow

The Scripps Research Institute, La Jolla, California

\$75,000 – 1-year grant

(September 15, 2016 – September 14, 2017)

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

Florian Eichler, MD, Principal Investigator

Sherman Alexander, MSc, Co-Principal Investigator

Massachusetts General Hospital, Boston, Massachusetts

\$96,128 – 1-year grant

(September 1, 2016 – August 31, 2017)

MECHANISM AND PATHOPHYSIOLOGICAL SIGNIFICANCES OF A GENETIC INTERACTION OF CYSTINOSIN

Bruno Gasnier, PhD, Mentor

Yann Terres, MSc, Research Fellow

University Paris Descartes, Paris, France

\$225,000 – 3-year grant

(September 1, 2016 – August 31, 2019)

PROFILING METABOLIC DYSFUNCTION CAUSED BY CYSTINOSIS

Norbert Perrimon, PhD, Mentor

Patrick Jouandin, PhD, Research Fellow

Harvard Medical School, Boston, Massachusetts

\$225,000 – 3-year grant

(September 1, 2016 – August 31, 2019)

KIDNEY ORGANOID: A NEW MODEL TO STUDY CYSTINOSIS

Alan Davidson, PhD, Principal Investigator

Teresa Holm, MD, PhD, Co-Principal Investigator

The University of Auckland, New Zealand

\$145,852 – 2-year grant

(September 1, 2016 – August 31, 2018)

DEVELOPMENT OF A TOPICAL, CONTROLLED RELEASE CYSTEAMINE EYE DROP

Morgan Fedorchak, PhD, Principal Investigator

Kanwal Nischal, MD, FRCO, Co-Principal Investigator

University of Pittsburgh School of Medicine, Pittsburgh, Pennsylvania

\$155,484 – 1-year grant

(September 1, 2016 – August 31, 2017)

ELUCIDATION OF ALTERED METABOLISM AND BIOMARKERS IN CYSTINOSIS USING LARGE-SCALE METABOLOMICS APPROACHES

Ilya Gertsman, PhD, Principal Investigator

Bruce Barshop, MD, PhD, Co-Principal Investigator

University of California, San Diego, California

\$160,589 – 2-year grant

(September 1, 2016 – August 31, 2018)

NLRP2 IN THE PATHOGENESIS OF CYSTINOSIS

Giusi Prencipe, PhD, Principal Investigator

Fabrizio De Benedetti, MD, PhD,

Co-Principal Investigator

Bambino Gesù Children's Hospital, Rome, Italy

\$180,400 – 2-year grant

(January 1, 2017 – December 31, 2018)

ACCELERATED RESEARCH GRANT PROGRAM

ONE YEAR STEM CELL RESEARCH ASSOCIATE SUPPORT

Stéphanie Cherqui, PhD, Principal Investigator

University of California, San Diego, California

\$141,081.60 – 1-year grant (July 18, 2016 – July 17, 2017)

Equipment and Services:

QX200 Droplet Digital PCR System - \$88,148.90

Cellometer Auto T4 Plus SK-150 - \$5,810

Two Macintosh computers - \$3,222.70

Transgenic mouse model to advance stem cell research - \$43,900

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Characterization of mTORC1 signaling in early pathogenesis of cystinosis

Corinne Antignac, MD, PhD, *Principal Investigator*

IMAGINE INSTITUTE (INSERM U1163), PARIS, FRANCE

OBJECTIVE/RATIONALE:

We recently reported that cystinosin, in addition to its cystine transport role, participates in amino-acid sufficiency signaling to the master cell growth regulator mTORC1. Additionally, it has recently been shown that the translation of mitochondrial components encoded in the nuclear genome is regulated by mTORC1. Oxidative stress and misshaped mitochondria are a central feature of cystinosis and our transcriptomic data suggest that young *Ctns* $-/-$ mice of FVB genetic background that do not develop Fanconi syndrome slightly upregulate a whole subset of mRNAs involved mRNA translation and in mitochondrial function. We therefore further study the mTORC1-related role of cystinosin in the early pathogenesis of cystinosis with a focus on one of its most important effector pathways that is mRNA translation.

PROJECT DESCRIPTION:

Our project for the next two years is to further characterize mTORC1 signaling and to analyze early defects in the translation of mRNA, especially those involved in mitochondrial function, under the control of the mTORC1/4E-BP1 axis in our animal and cellular models of cystinosis. We will reactivate mTORC1 signaling with amino acids after nutrient shortage and compare the subset of actively translated mRNAs (i.e. bound to ribosomes) between *Ctns* $-/-$ and $+/+$ mice kidneys. In the second aim of the project, we will as well further dissect the protein-protein interaction changes taking place in the mTORC1 activation cascade in knockout and CRISPR/Cas9 mutated cells lines in order to characterize the core mechanism of the mTORC1-signaling role of cystinosin.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

If our hypothesis turns out to be true, it will help to unravel the mechanisms of the strong and deleterious oxidative stress taking place in the proximal tubule in cystinosis. The implication of mRNA translation pathways and/or specific changes in protein-protein interactions in the mTORC1 cascade could further provide therapeutic candidate targets to counteract the down-regulation of mTORC1 signaling in the proximal tubule.

ANTICIPATED OUTCOME:

We expect to observe differences in translated mRNA involved in mitochondrial function upon mTORC1 reactivation by amino acids. On the second aspect of the project, we expect to observe that the lack of or deleterious mutations in cystinosin alter the protein-protein interactions changes taking place at the lysosomal membrane in the course of mTORC1 reactivation by amino acids. Finally, we will also continue using the expertise we developed in CRISPR/Cas9-mediated genome editing and obtain new cellular models that will be useful for the cystinosis research community.

Evaluation of $Ctns^{-/-}$ mice protection by oral supplementation with basic amino-acids: focus on kidneys

Pierre J. Courtoy, MD, PhD, *Principal Investigator*

DE DUVE INSTITUTE, BRUSSELS, BELGIUM

OBJECTIVE/RATIONALE:

Kidney proximal tubular cells (PTCs) are an early target of cystinosis and are not efficiently protected by cysteamine. The major source of the disulfide amino acid, cystine, in PTCs is via endocytic uptake of ultrafiltrated plasma proteins bearing disulfide bridges. In cystinotic mice, suppression of endocytic uptake by genetic ablation of the endocytic receptor, megalin, prevents cystine accumulation and protects PTCs. In normal cultured PTCs, basic amino acids block megalin. This project will test whether oral supplementation of cystinotic mice by basic amino acids also prevents cystine accumulation and protects PTCs.

PROJECT DESCRIPTION:

Cystinotic versus control mice will be fed after two months of age by food pellets containing a five-fold excess over the normal supply of either of two key basic amino acids, lysine and arginine, or the same amount of arginine precursor, citrulline. Mice will be regularly surveyed, monitored for kidney function and sacrificed at 6 or 9 months of age. Kidney structure, cystine content and crystals, endocytic function and expression of established tissue markers of the Fanconi syndrome (apical endocytic receptors and major solute transporters) will be evaluated as in our previous grants. Multiple organ samples will be collected for further studies (justified if a kidney benefit is documented). In parallel, the inhibitory power of individual basic amino acids, or their combination, and their exact mode of action will be determined in cultured PTCs.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This project is a translational therapeutic research. It is based on our recent discovery that a laboratory procedure, generic ablation in PTCs of a crucial endocytic receptor, megalin, offers spectacular protection against cystine accumulation, crystal formation and structural lesions. Acute treatment of normal PTCs with basic amino acids is known to block megalin. We will establish if chronic treatment of cystinotic PTCs is also effective, and protective.

ANTICIPATED OUTCOME:

There is good hope that sustained oral supplementation by basic amino acids will be well-tolerated by cystinotic mice and will significantly prevent PTC cystine storage, lesions and dysfunction. Such a finding would pave the way to rational dietary intervention against cystinosis in humans.

Alternative therapies for nephropathic cystinosis

Francesco Emma, MD, *Principal Investigator*

Laura Rita Rega, PhD, *Co-Principal Investigator*

BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

OBJECTIVE/RATIONALE:

We recently demonstrated that genistein, a natural compound enriched in soy and belonging to flavonoid compounds, is able to decrease cystine levels in cells from cystinotic patients. Moreover, we recently identified a second compound that shares similarities with genistein (hereby indicated as compound #2) that corrects cell defects of cystinotic cells in culture that are not corrected by cysteamine. Both genistein and compound #2 have good safety profiles and are efficient in culture at concentrations that are similar to those obtained in humans treated with these drugs.

PROJECT DESCRIPTION:

Our project is focused on testing new potential compounds for the treatment of nephropathic cystinosis, by analyzing their mechanisms of action, we will try to increase our understanding of the disease. To this end, we will first treat cystinotic mice with genistein or compound #2, with or without concomitant cysteamine therapy, to confirm in animal models the efficacy of these compounds and check for their toxicities. Secondly, we will screen other compounds known to be similar to genistein and to compound #2, for their ability to rescue metabolic pathways that are altered in cystinotic cells but are not rescued by cysteamine. Finally, we will perform experiments aimed at clarifying cellular mechanisms by which selected drugs ameliorate the cystinotic cell phenotype.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS - ANTICIPATED OUTCOME:

Our project will test the potential therapeutic effects and safety profile of new drugs in animal models of nephropathic cystinosis (mice). These experiments are aimed at finding alternative or complementary therapies to cysteamine, which cannot cure all aspects of nephropathic cystinosis. We are also hopeful to identify new agents that may be more potent, more suitable or less toxic than the ones that we have already identified. By analyzing mechanisms by which these compounds correct the cell defects of cells obtained from patients with nephropathic cystinosis, we hope to increase our understanding of the disease.

Biophysical study of cystinosin and PQLC2

Bruno Gasnier, PhD, *Research Mentor*

Rossella Conti, PhD, *Research Fellow*

PARIS DESCARTES UNIVERSITY, FRANCE

OBJECTIVE/RATIONALE:

This project focuses on cystinosin, the lysosomal cystine transporter defective in cystinosis, and on another lysosomal amino acid transporter, termed PQLC2, which underlies cysteamine therapy of cystinosis. We aim to understand better their functional properties, with potential impact on our molecular understanding of cystinosis and its current drug treatment.

PROJECT DESCRIPTION:

Membrane transporters such as cystinosin and PQLC2 operate through an intricate cycle of conformational changes. In other words, they constantly change their shape. We will use a combination of molecular biological, biochemical and biophysical techniques to dissect out these conformational cycles. For cystinosin, we will try to block the conformational changes and trap the protein in a specific state to help structural studies by another laboratory. We will also study how the internal acidity of the lysosome, a powerful energy source for diverse lysosomal processes, drives cystine export by cystinosin. For PQLC2, we will investigate how another amino acid, arginine, interferes with its transport activity.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

The biophysical study of cystinosis may help understand the molecular effect of some patient mutations. The identification of artificial mutants trapped in a defined conformation should help dissecting novel cystinosin functions, unrelated to the cystine transport function, reported by other laboratories. The biophysical study of PQLC2 will tell whether variations in cellular levels of arginine might interfere with the cystine-depleting action of cysteamine.

ANTICIPATED OUTCOME:

We expect to get a better understanding of the molecular and cellular biology of cystinosin and of the molecular effect of cysteamine on patient lysosomes. Our research should also provide molecular tools (cystinosin or PQLC2 mutants) useful for the cystinosis research community.

Pharmacokinetic evaluation and optimization of cysteamine precursors

Vincent Stanton, Jr., MD, *Principal Investigator*

Patrice Rioux, MD, PhD, *Co-Principal Investigator*

THIOGENESIS THERAPEUTICS, INC., SAN DIEGO, CALIFORNIA

OBJECTIVE/RATIONALE:

Cysteamine is rapidly metabolized in the body, which requires frequent administration of high doses to maintain therapeutic levels. The dosing schedule is inconvenient, and because cysteamine is rapidly absorbed, peak drug levels are high, causing unpleasant side effects. This research will test the properties of several cysteamine precursors in laboratory animals. The precursors were designed to be slowly converted into cysteamine in the intestine. They are expected to reduce the high peak cysteamine concentrations associated with side effects, while extending the interval between doses.

PROJECT DESCRIPTION:

Cysteamine precursors will be administered to rats. The absorption of cysteamine into the blood, distribution into tissues, and metabolism and elimination will be studied. The pharmacokinetic properties of the cysteamine precursors will be compared to the properties of cysteamine itself. The effect of co-administered food or pharmacological modulators of cysteamine precursor degradation and absorption will also be studied. The best performing cysteamine precursor from the rat experiments will then be studied in more detail in mini-pigs, a species whose gastrointestinal anatomy and physiology more closely resemble that of humans. The effect of drug formulation on cysteamine precursor pharmacokinetics will also be investigated in mini-pigs. These studies are expected to allow selection of a cysteamine precursor and formulation for subsequent human studies.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Cysteamine, while not a cure, is an effective therapy for cystinosis when administered at the recommended dose and schedule. However, both the dose and schedule are challenging to adhere to, particularly because of the side effects associated with peak blood levels of cysteamine. This research is aimed at characterizing several compounds designed to address these shortcomings by limiting the rise in blood cysteamine concentration following a dose, and by extending the time over which cysteamine is available for absorption from the gut.

ANTICIPATED OUTCOME:

This research will show whether cysteamine precursors can produce sustained cysteamine blood levels in the therapeutic range in laboratory animals, in contrast to the sharp, high peak concentration and subsequent rapid decline characteristic of the two presently marketed formulations. Several methods for modulating the breakdown of cysteamine precursors and the absorption of resulting cysteamine will also be studied, providing further understanding of how to optimize and individualize dosing of cysteamine precursors.

Lessons from the fruit fly: How cells cope with cystinosis deficiency

Matias Simons, MD, Principal Investigator

IMAGINE INSTITUTE, PARIS, FRANCE

OBJECTIVE/RATIONALE:

We are using the fruit fly *Drosophila melanogaster* to understand the physiological role of the lysosomal cystine transporter cystinosis (*CTNS*) in humans. We deleted and modified the gene *CTNS* on the genomic level and observed several whole-animal and cell-specific alterations that partially overlap with the clinical picture and disease progression of human cystinosis patients. We are particularly interested in understanding the metabolic changes that occur in cells with mutated *CTNS* and search for ways to correct this imbalance via modifications in food and drug treatment or at the level of gene regulation.

PROJECT DESCRIPTION:

We have already deleted or tagged *CTNS* in the fly genome and re-inserted modified versions of fly or human *CTNS* (with mutations and fluorescence markers) under their own regulatory gene expression elements (first funding period 2014-2016). This will further allow us to study cellular perturbations in the absence of *CTNS* and the behavior of fluorescently labelled *CTNS*. By using a combination of state-of-the-art high-throughput bioanalytical methods and high-resolution microscopy, we aim to understand the alterations on gene expression, growth signaling and metabolism that are caused by *CTNS* depletion on the cellular and the organismal level. With this setup, we expect rapid progress in the understanding of disease mechanisms of cystinosis.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

We hypothesize that amino acids and other catabolic products produced by the lysosome can activate signaling pathways that are specific to the homeostasis of certain cell types, e.g. proximal tubular cells. The combination of transcriptome and metabolome analysis will allow the identification of relevant pathways and gene targets which can be targeted for development of novel therapies. We will use the fruit fly as an *in vivo*-eukaryotic system to screen for genes and substances that could lead to the rescue of developmental and life-reducing phenotypes caused by *CTNS* deficiency.

ANTICIPATED OUTCOME:

We are focusing on novel metabolic pathways that account for the observed phenotypes of the *CTNS* mutant fly. To accelerate the outcome, we are collaborating with the group of N. Perrimon (Harvard Medical School, CRF grant 2016), that also uses the fruit fly to study cystinosis. Together, we hope to unravel the metabolic alterations caused by *CTNS* deficiency and to identify food supplementations or genetic manipulations that can rescue *CTNS*-dependent phenotypes.

Sleep disturbance and memory function in Nephropathic Cystinosis

Doris Trauner, MD, *Principal Investigator*

UNIVERSITY OF CALIFORNIA, SAN DIEGO

OBJECTIVE/RATIONALE:

Difficulty with memory may be a problem in adults with cystinosis. Many adults also complain of difficulty with sleep. Sleep disturbance can be associated with memory problems. The objective of this study is to identify sleep disturbances in adults with cystinosis and to determine whether memory difficulties are more likely to occur in people with sleep disorders. Results could positively impact the quality of life in adults with cystinosis by identifying possible interventions that could reduce memory problems.

PROJECT DESCRIPTION:

Adults with cystinosis will be invited to come to San Diego for a one-night, non-invasive sleep study in a sleep laboratory at UCSD Medical Center. On the day of the sleep study, a number of cognitive tests will be administered to the participant to evaluate attention, memory and general cognitive functioning. Information from the cognitive testing and the sleep study will then be analyzed to determine whether there is an association between sleep disturbance and memory problems in people with cystinosis.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This study may lead to better understanding of why some adults with cystinosis experience problems with their memory, and could lead to treatments for sleep disturbance that could improve memory in those individuals, thus improving the quality of life for adults with cystinosis.

ANTICIPATED OUTCOME:

We hope to learn how frequently adults with cystinosis experience memory problems and sleep disturbance, and whether the two are related. The results could potentially lead to treatments for adults with memory difficulties and/or sleep problems.

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 14 short years. Since its inception, CRF has raised more than \$39 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 \$3 million was available for the 2017 spring call for research proposals and fellowship grants. The grant awards will be announced in June 2017. Details and guidelines for applications are available online at the CRF website: www.cystinosisresearch.org/research/for-researchers.

In 2016, CRF issued 17 new grants in five countries totaling \$ 2.79 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 (See page 72). 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 the 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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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 \$39 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.



WITH YOUR **courage** KINDLED

YOU STEP ONTO NEW GROUND

YOUR EYES YOUNG AGAIN

WITH ENERGY TO **dream**

A PATH OF PLENITUDE OPENS BEFORE YOU
